SPECTRUM OF THALASSEMIA AND HEMOGLOBINOPATHIES IN A TERTIARY CARE HOSPITAL

A THESIS SUBMITTED TO THE BABA FARID UNIVERSITY OF HEALTH SCIENCES, FARIDKOT (PUNJAB)

FOR THE AWARD OF

MD

IN

PATHOLOGY

BY

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Dedicated

to

my Family

&

Teachers

ACKNOWLEDGEMENT

"As we express our gratitude, we must never forget that the highest appreciation is not to utter words, but to live by them."

First of all, I humbly bow my head to the Almighty God for the strength, endurance and perseverance. I have always felt his presence in the form of my respected teachers while sailing through high and low challenges of my career, thus moving successfully towards my destination and serving the purpose dutifully that I am here for.

Any research project is a challenging task and a research worker could accept it with better courage, and work on it with true dedication when he is fortunate enough to continuously receive inspirational blessings of eminent and patronizing teachers and guides. The culmination of this onerous endeavour would not have been possible short of unflinching and spirited support I have received all along the way.

Words cannot express my deepest gratitude and indebtness to my revered guide and mentor **Dr. Bhavna Malhotra**, Professor, Department of Pathology, Dayanand Medical College and Hospital, Ludhiana whose limitless efforts and keen interest helped me throughout this study. I thank her for her patience and timely advice, without which I have floundered in this study. She is an icon under whose vision and dedication, many students have reached the pinnacle of their careers. She provided the best encouragement, facilities and motherly affection that helped me to overcome whatever obstacles came during the course of my study. I will always be proud of the fact that once, I was her student.

A debt so precious as that of learning and knowledge cannot be repaid except through gratitude. By providing constructive criticism, It is indeed my great privilage and earnest duty to register my humble and respectful gratitude towards my worthy, experienced and esteemed teacher, **Dr. Vikram Narang**,

Associate Professor, Department of Pathology , Dayanand Medical college and Hospital, Ludhiana. It has been a great privilege to work with a pedagogue of his outstanding caliber and vast knowledge. He handed my unending but seemingly trivial problems with great fineness. I am grateful to him for accommodating me with great patience at all times.

With great regard and due decorum I acknowledge my sincere thanks to **Dr. Harpreet Kaur**, Professor and Head, Department of Pathology, Dayanand Medical College and Hospital, Ludhiana, for her invaluable advice and guidance throughout the course of the study.

It is with supreme sincerity and deep sense of gratitude I thank my esteemed teacher **Dr. Neena Sood,** Former Head and Professor, Department of Pathology, Dayanand Medical college and Hospital, Ludhiana for her guiding wisdom which many a times supported my sagging spirits when face with a multitude of hurdles. I feel deeply indebted to her for her untiring help in the preparation of this dream work of mine and helping me to prepare for the future.

I take this opportunity to convey my heartfelt gratitude and sincere thanks to **Dr. Pavneet kaur, Dr. Aminder Singh, Dr. Sumit Grover and Dr. Ruchita Tyagi,** Department of Pathology, Dayanand Medical college and Hospital, Ludhiana who have been a source of inspiration and helped me throughout my post graduate training at DMCH.

I am Indebted to my parents**, Mrs Ritu Jain** and my uncle **Mr. Munish Jain** for their valuable support. Without their silent blessings and encouragement this study would never have seen this day.

I am highly thankful to my brother, **Mr. Mayank Jain** for his unending support and help.

I am thankful to the members of hematology **Mr. Subash Chander Sharma and Mrs Haripriya.** Technical staff of Immunopathology especially **Mr. Kapil and Mr. Vishal Sharma** of Department of Pathology for their skilled assistance and cooperation.

I take this opportunity to thank my colleagues **Dr.Navita, Dr.Shefali, Dr.Aastha and Dr. Harsimran** for sharing the highs and lows of residency and life.

I extend my sincere thanks to **Dr. Ankita Soni, Dr. Saveena Jindal**, my seniors and juniors especially **Dr. Rashi and Dr. Ramneek** for their valuable support and helping me in my daily work. I am also thankful to the department staff for the unending help during this study.

I acknowledge the help of all those invisible hands who are responsible for the successful outcome of the dissertation.

"My teacher How Can I Ever Thank You Enough, What You Have Given Me Will Stay with Me a Life Time"

DR ANSHUL JAIN

LIST OF ABBREVIATIONS

BTT	Beta-thalassemia trait
BP	Base pair
CBC	Complete Blood Count
DEL	Deletion
DLC	Differential Leucocyte Count
DMCH	Dayanand Medical College and Hospital
DNA	Deoxy Ribonucleic Acid
EDTA	Ethylene Diamine Tetra Acetic Acid
Fl	Femtoliters
g/dl	Grams per deciliter
Hb	Hemoglobin
HPFH	Hereditary persistence of Fetal Hemoglobin
HPLC	High Performance Liquid Chromatography
IEF	Isoelectric Focussing

IVS	Intervening Sequence
MCH	Mean corpuscular Hemoglobin
MCHC	Mean Corpuscular Hemoglobin Concentration
MCV	Mean Corpuscular Volume
PBS	Peripheral Blood Smear
Pg	Pico Grams
RBC	Red Blood Cell
RDW	Red Cell Distribution Width
TLC	Total Leucocyte Count
WHO	World Health Organization

ABSTRACT

Introduction: Thalassemia and hemoglobinopathies are common hematologic disorders causing significant health care burden on society. The disease incidence vary among populations of different regions in diverse country like India.

Aims and objectives: To study Spectrum of thalassemia and hemoglobinopathy.

Material and methods: The present 2.5 year study was conducted in Department of Pathology, Dayanand Medical College and Hospital where all sample received for HPLC were analysed with respect to clinical findings, hematologic parameters and HPLC chromatograms and cases diagnosed with thalassemia or hemoglobinopathy were included.

Results: Of the 405 cases included majority were in age group of 21 to 30 years (181 cases, 44.5%) with female preponderance (M:F=1:1.9) Beta-thalassemia trait (63%) was the commonest followed by HbD Punjab heterozygous (9.3%), Beta-thalassemia homozygous (5.4%), HbQ India heterozygous (1.5%), Compound heterozygous for HbD Punjab and Beta thalassemia (1%) and Sickle cell heterozygous, Sickle cell homozygous, HbE heterozygous and HbD Iran heterozygous and compound heterozygous for HbQ and β thalassemia (0.5% each). Patients with borderline HbA2 constitute 65 cases (16.1%).

Conclusion: Thalassemia and hemoglobinopathies can be easily diagnosed using hematologic parameters and HPLC. Establishing early antenatal diagnosis along with family screening and molecular characterization is essential.

Keywords: Thalassemia, Hemoglobinopathy, HPLC, Molecular genetics

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KEY TO MASTER CHART

RBC- Red Blood Cell

Hb- Hemoglobin

MCV- Mean Corpuscular Volume

MCH- Mean Corpuscular Hemoglobin

MCHC- Mean Corpuscular Hemoglobin Concentration

RDW- Red Cell Distribution Width

INTRODUCTION

Hemoglobinopathies are a group of autosomal recessive inherited hemoglobin diseases, defined by qualitative (variant Hbs) and quantitative (thalassemia syndrome) abnormalities affecting the globin chains.[1] They are classified into two broad groups, the hemoglobinopathies and the thalassemia. The hemoglobinopathies are characterized by the production of structurally defective hemoglobin due to abnormalities in the formation of the globin moiety of the molecule.[2] The thalassemia are characterized by either the reduction or the absence of synthesis of α- or β-globin chains, known as the α-thalassemia and β-thalassemia respectively.[1] The clinical spectrum of these disorders varies from asymptomatic conditions to serious disorders like thalassemia major that require regular blood transfusions, widespread medical care[3,4] and bone marrow transplantation is the only curative option available. These inherited disorders are major public health problem in the world including India.[5]

The β-thalassemia and their interaction with structural Hb variants, beta HbS, HbE and HbD are a major public health problem in India. Some of the less commonly found Hb variants in India that are detected during screening programs are Hb Lepore, HbD Iran, HbJ Meerut, HbH disease and HbQ India.[6] The distribution of specific disorders varies geographically and by community. WHO estimated that 7% of the world population is a carrier for Hb disorders.[7] The cumulative gene frequency of hemoglobinopathies in India is 4.2% with population of 1.25 billion. Every year 10,000 children with thalassemia major are born in India, which constitutes 10% of the total number in the world.[8] The frequency range of beta-thalassemia is 3.5% to 15% in Indian population.[9] Accurate and timely detection of various Hb variants including β-thalassemia trait can prevent the occurrence of more serious disorders like thalassemia major in newborns.[10] Each year about 3,00,000 infants are born with a major hemoglobinopathy.[10,11,12]

In some of the developing countries individuals with thalassemia major are either untreated or transfused inadequately. Persistent severe

anemia leads to various consequences. The common findings in such underprivileged children include growth retardation, pallor, jaundice, poor musculature, hepatosplenomegaly, leg ulcers, and development of masses from extramedullary hematopoiesis and skeletal changes that result from expansion of the bone marrow.

On the other hand, complications due to iron overload contribute to the major issues in those patients receiving regular transfusion. The iron overload leads to serious complications including endocrine complication (growth retardation, diabetes mellitus and insufficiency of the parathyroid, thyroid, pituitary, failure of sexual maturation and less commonly dilated myocardiopathy, liver fibrosis and cirrhosis.

The extremely high frequency of the Hb disorders compared with other monogenic diseases reflects several other different processes. First, and foremost, is natural selection in which carriers for these diseases have been protected during evolution from severe malaria. Hence they have lived longer in malaria endemic areas and produced more children until the numbers are balanced by the loss from the community of homozygotes. A second critical factor is the high frequency of consanguineous marriages which tend to increase the frequency of any Mendelian recessive disorder. Additional factors that maintain the high gene frequency include founder effects and gene drift.[13]

More than 200 mutations in β globin genes have been identified to cause the disease. The observed clinical heterogeneity in development of β thalassemia led to the identification of more than 200 mutations so far. The clinical spectrum displayed by the thalassemic individuals is dominantly influenced by the type of mutation inherited.

Thalassemia and other hemoglobinopathies can cause life threatening situations and chronic ill health. They pose economical and psychological burden on the affected individual, his family and country as a whole. Hence a detailed study needs to be conducted and steps should be taken to improve the quality of life of these patients and to prevent it by population screening, genetic counselling, prenatal diagnosis and premarital counselling as well. Almost 10 billion rupees per year is spent for thalassemia patients in India. In India due to

sociocultural practices, these disorders are restricted to particular geographic areas, tribes and religion. Early marriages among individuals of the same caste or in close blood relations makes it to know the spectrum of thalassemia and other hemoglobinopathies. All it takes is a simple blood test to identify silent carriers so as to avoid marriages between them.[14]

AIMS AND OBJECTIVES

To study Spectrum of Thalassemia and Hemoglobinopathies.

REVIEW OF LITERATURE

Hereditary disorders of hemoglobin result either from quantitative defect (reduced or absent synthesis of one or more of normal globin polypeptide chain) known as Thalassemia or qualitative defect (structural alteration of globin polypeptide chain) known as hemoglobinopathies. Alpha and beta-thalassemia are the commonest single gene Hb disorders in the world.[15-18] The alpha and beta chains of hemoglobin are synthesized independently under separate genetic control and in the normal state, synthesis of the two chains is balanced.[19] Hemoglobinopathies occur due to mutations, like Hb-Lepore, HbD-Punjab and HbE or failure to complete the normal neonatal switch from HbF to HbA, referred to as hereditary persistence of fetal hemoglobin (HPFH).[20,21]

Both thalassemia and hemoglobinopathies are autosomal recessive inherited disorders primarily affects globin moiety of Hb molecule.[22] Thalassemia occurs particularly in high frequency in broad belt from Mediterranean basin through the Middle East, Indian subcontinent, Burma, Southeast Asia, Melanesia and the islands of the pacific. [23]

According to recent data collected through the Hereditary Disease Program of the World Health Organization (WHO) and based on local surveys and reports by visiting experts, the carriers of hemoglobin disorders in the world are estimated to be 269 million which constitutes 4.5% of the world population and constitutes 30 million people from India.[13]

The carrier rate for beta thalassemia gene ranging from 1 to 3% in Southern India and 3 to 15% in Northern India.[24] On an average 1 in every 25 Indian is a carrier of thalassemia and it is estimated that there are about 65,000-67,000 beta thalassemia patients with more than 12,000 infants born each year with clinically significant hemoglobinopathies.[25,26]

An estimated 300,000 babies are born each year with severe inherited disease of hemoglobin and that over 80% of these births occurs in low or middle income countries. As these countries go through the

epidemiological transition which is characterized by a reduction in childhood and infant mortality due to improved public health measures, the infants who previously died of these disorders before they were recognized, are now surviving due to early diagnosis and treatment. So hemoglobinopathies will constitute an increasing global health burden.

There is limited data available about the true frequency, natural history and survival of patients with these disorders, the information is absolutely critical in providing information to Government and International health agencies about the true global burden of these disorders.[13,27]

The carrier state for Beta-thalassemia in India varies from 1% to 17% with an average of 3.2%.[28,29] Beta-thalassemia trait is most common in communities such as Sindhi, Parsee, Lohar and different ethnic groups of Punjab, Bengal and Gujarat.[30] Worldwide Sickle cell Hb is the most common pathological Hb variant. The presence of gene for Sickle cell Hb was first detected in Nilgiri hills of the Southern part of India.[30] HbE-Beta thalassemia with HbS are also very common problem with frequency reported from different parts of India.[31-32] HbD-Punjab is one of the most commonly encountered abnormal hemoglobin in the world[33] and has a high frequency in North West India mainly Punjab with a incidence of 2-3%.[34,35] It is well established that incidence of HbE gene in north eastern region of India is one of highest in world. Different states in north eastern region show variable incidence of HbE varying from 16.2% to 47.3%.

Historical Background

Beta-thalassemia is a monogenic hematological disorder first discovered by Thomas Cooley and Pear Lee[36] (Cooley & Lee, 1925), later termed as Cooley's anemia or Mediterranean anemia. George H.Whipple and William L. Brad ford[37] (1932) described the pathology of the condition and they first used the term 'thalassemia' in their classical paper in 1932. The word thalassemia is derived from the Greek word 'thalassa', which means 'the sea' because early patients were all from regions

surrounding the Meditteranean area and “emia” means related to blood.[21]

The first description of clinical features of various types of thalassemia were published during the period between 1925 and 1940. The pattern of genetic inheritance of thalassemia was first described during the period of 1940–1950. Valentine and Neel[38] (1944, 1948) classified Cooley’s anemia based on the severity of the disease naming the milder forms as thalassemia minor for heterozygote state and severe type as thalassemia major. They have done pioneer work on the genetic transmission of thalassemia.

In 1946 Vecchio[39] noticed that the hemoglobin of Cooley’s anemia patients was found to be more alkali-resistant than normal adult hemoglobin. Thereby he suggested that the amount of fetal hemoglobin is more in patients with Cooley’s anemia than what is usually present after first year of life. The microcytic anemia of Cooley’s disease is often accompanied by the presence of large, pale macrocytes and target cells. Dameshek pointed out this phenomenon and called the condition as ‘target-cell anemia’ or ‘leptocytosis’.[40]

In 1952 Rich[41] suggested that thalassemia results due a defect in HbA synthesis with persistent production of HbF. The important theoretical model for the genetic basis of thalassemia was set out by Ingram and Stretton[42] in 1952. In 1981, Niazi Coleman and Hoeffler introduced chorionic villous sampling which provides sufficient DNA for testing.[43] In 1982, Thomas and his co-workers[44] first documented bone marrow transplantation for thalassemia. In 1983, Alter introduced electrophoresis and Isoelectric focussing (IEF) for globin chain separation.[44] In 1991, NY Varawalla et al studied the spectrum of Beta-thalassemia in Indian subcontinent which form the basis for prenatal diagnosis.[45]

Structure of Hemoglobin

Normal adults have a major hemoglobin called HbA, comprising about 97% of adult red cells[2] and a minor component HbA2 which accounts for 2–3%. The main hemoglobin in fetal life is HbF, traces of which are

found in normal adults. Each consists of two separate pairs of identical globin chains. In HbA, alpha chain combine with beta- chain ($\alpha 2\ \beta 2$), In HbA2, alpha chain combine with delta chains ($\alpha 2\ \delta 2$) and in HbF, alpha chain combine with gamma chain. ($\alpha 2\ \gamma 2$)

The primary structure of hemoglobin is the four polypeptide chains attached to heme molecule. Each chain is arranged in a series of eight helical segments joined by short non-helical segments, referred as secondary structure. Each heme molecule is enclosed in a pocket by the folds of the chain.

Globin gene Clusters

The alpha gene cluster usually contains one functional ε gene and two alpha gene ($\alpha 1\ \alpha 2$). The alpha gene cluster also contains four pseudogenes $\psi\varepsilon$, $\psi\varepsilon$, $\psi\alpha$ and θ. Both the genes are located in a region of homology approximately 4 kb long and interrupted by two small non-homologous regions. Homologus regions are due to duplication of gene and non-homologus regions are due to DNA insertions into the non-coding regions around one of the two genes. The alpha gene cluster is located in the telomeric region of chromosome 16.

The β gene cluster is located in the short arm of chromosome 11, as G γ Aγ gene pairs and five functional genes $\varepsilon, G\gamma, A\gamma, \delta$ *and* β. Different hemoglobins are produced during development and two globin gene switches take place. The embryonic ε gene is found in the embryonic phase produces hemoglobin - Hb Gower 1 ($\varepsilon 2\ \xi 2$) and Hb Gower 2 ($\alpha 2\ \varepsilon 2$) . The duplicated fetal γ genes encode the globin chains Gγ *and* $A\gamma$ in the polypeptide chain, the fetal Hemoglobin contains $\alpha 2\gamma 2$ chain and the fetal to adult hemoglobin switch occurs during the perinatal period. (Fig. 1)

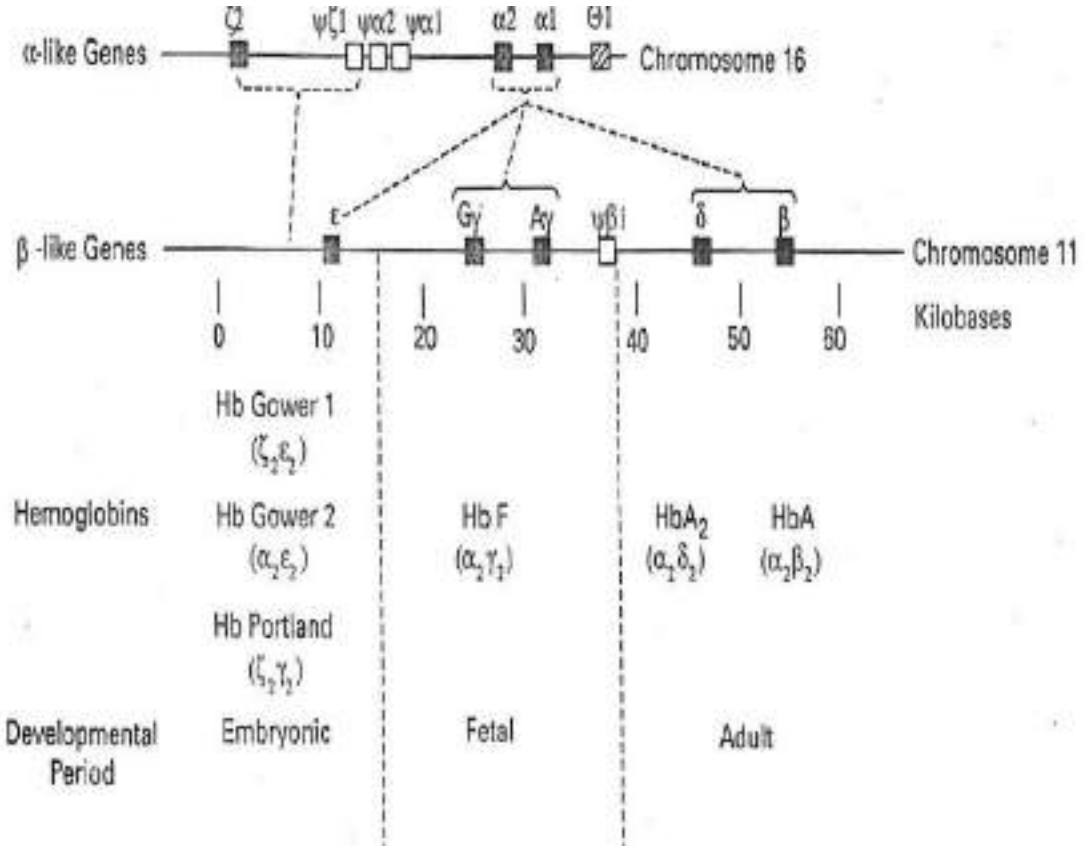

Fig 1: Chromosomal Organization of the globin genes and their expression during development.

Classification of Thalassemia

Alpha thalassemia and Beta thalassemia exist in both the homozygous and heterozygous states and the genes for thalassemia may interact with those of the hemoglobin structural variants. Concurrent presence of alpha thalassemia modifies the phenotype of beta thalassemia. Alpha thalassemia is not a significant problem in India because the severe alpha deletion mutations are less common in this region and is clinically silent but is widely prevalent in tribal population. The alpha thalassemia is primarily due to gene deletion directly causing reduced alpha globin chain synthesis. Beta thalassemia is usually caused by point mutation rather than large deletion. (Table 1)

Table 1: Classification of Thalassemia syndromes[46]

Clinical Syndromes	Genotypes	Clinical Features
1)β thalassemia		
β thalassemia major	(β^0/β^0, β^+/β^+, β^0/β^+)	Severe; Blood transfusion dependent
Beta thalassemia intermedia	(β^0/β^+, β^+/β^+, β^0/β, β^+/β)	Severe but does not require regular blood transfusions
β thalassemia minor	(β0/β, β+/β)	Asymptomatic with mild or absent anemia; red cell abnormalities seen
2)α thalassemia		
Silent carrier	-/α α/α	Asymptomatic no red cell abnormality
α thalassemia trait	-/-α/α -/α-/α	Asymptomatic red cell abnormalities seen like β thalassemia minor
HbH disease	-/- -/α	Severe; resembles β thalassemia intermedia
Hydrops fetalis	-/- -/-	Lethal in utero without transfusions

a) **Thalassemia Major:** Homozygous state, most severe form, characterized by severe anemia, high HbF levels and is transfusion dependant.

b) **Thalassemia Intermedia:** Milder form of thalassemia major. It is moderately severe in hemolysis. Patients grow into adulthood and are not transfusion dependant.

c) **Thalassemia Trait:** Is a heterozygous asymptomatic carrier state. The carrier does not exhibit any symptoms and leads an absolutely normal life. These descriptive term do not have a precise genetic meaning, they remain useful in clinical practice.[47]

Pathophysiology of Thalassemia Major

The basic defect in Beta thalassemia is a reduction in the β globin chain production. On the other hand, α globin chains are produced in excess leading to imbalance in the production of globin synthesis.

In all severe forms of Beta thalassemia there is persistence of HbF synthesis beyond birth and infancy but in variable degrees. Yet overall HbF synthesis and output is essentially inadequate to compensate for the marked deficiency of HbA. In other words there is never a match between the production of α chains to that of output of β and γ chains in Beta thalassemia. Thus the hallmark of this disease is the unbalanced production of globin chains and an excess of α chains is therefore the pathogenic marker of β thalassemia.

Classically in thalassemia, there is defect in the maturation of erythroid precursors along with ineffective erythropoiesis and a shortened red cell survival as the excessive unbound α chains precipitate within the red-cell precursors in the marrow and in their progeny in the peripheral blood. There is intense proliferative drive in the ineffective bone marrow by the resultant anemia, which leads to its expansion. Thereby a variety of growth and metabolic abnormalities develop and also results in array of skeletal deformities. There is exacerbation of the anemia undoubtedly due to shunting of blood through the massively expanded marrow together with splenomegaly.[48] The characteristic increased iron absorption and the consequent iron loading occurs due to the hyperplasia of the bone marrow which is often exaggerated by the need for regular packed cell transfusion. This leads to progressive iron deposition in various

tissues. If the excessive iron that gets deposited is not removed by any means, multi organ failure ensues that culminates to death of these patients.

In 1966, Fessas et al. described the presence of inclusion bodies in erythroblasts of thalassemic patients, suggesting that they were precipitates of α-chains. The composition of inclusion bodies in β-thalassemia, completely consisting of precipitated α-chains, has been recently confirmed by immunoelectron microscopy. Oxidation of excess α-chains results in the formation of hemichromes, whose basic structure consists of the covalent binding of distal histidine E7 to the sixth coordination site of the heme iron. Irreversible hemichromes and denatured α-chains precipitate as inclusion bodies early during differentiation and throughout erythroid maturation.[49]

α-Chain precipitation in the red cell membrane causes structural and functional alterations with changes in deformability, stability and red cell hydration. Protein 4.1, a major component of the cytoskeleton, undergoes partial oxidation in β-thalassemia resulting in its defective capability to mediate the formation of the spectrin–protein 4.1–actin complex, which is critical to maintain cytoskeleton stability.[49]

Red blood cells in β-thalassemia lose K+, store Ca2+ and are dehydrated, resulting in altered deformability. Besides oxidation, free α-chains are subjected to degradation, resulting in the formation of denatured α-globin protein, heme and free iron. Free iron, via the Fenton reaction, generates reactive oxygen species, which cause lipid and protein peroxidation with consequent damage to red cell membranes and intracellular organelles. Apoptosis could contribute significantly to ineffective erythropoiesis and occurs primarily at the polychromatophilic erythroblast stage.

The ineffective erythropoiesis and anemia has several consequences in the organism as a whole, producing the clinical picture of the disease. The first response to anemia is an increased production of erythropoietin, causing a marked erythroid hyperplasia. Anemia may produce cardiac enlargement and sometimes severe cardiac failure. Erythroid expansion produces skeletal deformities, osteoporosis and

occasionally extramedullary masses and contributes to splenomegaly. Untreated or undertreated thalassemia major patients have retarded growth as a result of anemia and the excessive metabolic burden imposed by erythroid expansion. Environmental factors such as poor nutrition and infections may contribute to growth failure. The high vascularization of expanded marrow results in an increase in plasma volume, which is associated with splenomegaly, aggravates the anemia. Ineffective erythropoiesis is associated with increased iron absorption, which in transfusion- dependent patients, is largely increased by the blood transfusions. The major cause of morbidity and mortality is the effect of iron deposition on the endocrine organs, liver and heart, which results from increased intestinal absorption and the effects of blood transfusion. Removal of the abnormal red blood cells by the reticuloendothelial elements of the spleen results in splenomegaly and hypersplenism, which when severe may exacerbate anemia and cause thrombocytopenia and leucopenia.[49]

The pathophysiology of the α-thalassemia is different because the excess β chains resulting from defective alpha-chain production form β4 molecules or hemoglobin H, which is soluble. However, hemoglobin H is unstable and precipitates in older red cells. Hence, the anemia of alpha-thalassemia is hemolytic rather than dyserythropoietic.[50]

Mutations in beta thalassemia major

More than 150 mutations causing Beta-thalassemia have been reported from different parts of the world.[51] Studies conducted in India have identified about 28 mutations in Indian population. Of these five to six mutations are found to be common.[52-54] These include IVS 1-5, 619 bp del, IVS 1-1, frameshift 8-9, 41-42 and codon 15. The predominant mutation in most of the states being IVS 1-5. Overall common mutations account for 90-93.6 percent.[54]

Most of these mutations cause defects in transcription, RNA splicing, RNA modification and translation because of frame shift and

nonsense codons or produce highly unstable β -globin products. However, vast majority of β -thalassemia syndromes are caused by point mutations within the β –globin gene itself or in its immediate flanking sequences.[55] The clinical spectrum displayed by the thalassemic individuals is dominantly influenced by the type of mutation inherited. Hence, diverse forms of thalassemia arise in mutations that affect nearly every step in globin chain expression i.e. transcription, translation, processing and so on.

Laboratory diagnosis[56]

a) **Complete Blood Count-** On peripheral smear there is anisopoikilocytosis with target cells, tear drop cells and nucleated RBCs with decreased red cell parameters, Hb is 3-8gm%, Reticulocyte count<2%, Mentzers index is <13.

b) **Iron status-** Serum Ferritin>1000ug/L and transferrin saturation is markedly increased, Total iron binding capacity is reduced and Serum iron is increased. Serum LDH is high and Serum haptoglobin is low.

c) **Bone marrow-** Hypercellular with erythroid hyperplasia and M:E ratio is reversed. Myelopoiesis and megakaryopoiesis is normal.

d) **HPLC-** Increased HbF levels and HbA2

f) **Mutation studies**- Provide information about specific mutation by DNA analysis.

Thalassemia Intermedia- Clinical entity in which patients have clinical spectrum in between Beta- thalassemia trait and thalassemia major, non-transfusion dependant phenotype. Modell and Berdoukas reported that only 11% of patients presented in first year of life, 30% presented in second year and 59% presented later in life.[57]

Clinical features

Anemia of varying degree and mild jaundice are present where growth and development remained normal. The spleen is palpable from the beginning in majority of patients and increases with time. Hypersplenism

is frequently heralded by decreasing Hb levels. thrombocytopenia and neutropenia then ensues.[57]

Laboratory Diagnosis

Hb is 7-8gm% shows moderate degree of anisopoikilocytosis with microcytic hypochromic cells, HbF is 10-30% and marrow iron stores are increased.

Thalassemia trait- heterozygotes for thalassemia gene and are asymptomatic. Women during pregnancy are diagnosed in thalassemia screening program. Prevalence varies from 1.0-14.9% in various regions of India. It is clinically important to differentiate it from Iron deficiency anemia because each has an entirely different cause, treatment and prognosis. Antenatal screening is the important step to identify women having the risk of producing child affected with hemoglobinopathy.

Laboratory Diagnosis

CBC- Hb-10-12gm%, RBC count is increased, MCV and MCH is reduced but MCHC is nearly normal. Mentzer index is <13 while >13 indicates Iron deficiency anemia.

On peripheral smear there is anisocytosis with microcytic and hypochromic RBCs and few target cells.

HPLC- HbA is 90-95% and an elevated HbA2 with an average value of 5% is characteristic of β-thalassemia trait.[58] Elevated HbA2 may be the only abnormality detectable by ordinary means, so normal HbA2 value does not mean that an individual cannot produce a child with severe homozygous thalassemic features.[59]

Thalassemia syndromes

a) HbS thalassemia syndrome- Double heterozygote state of HbS and Beta thalassemia. HbS is less than 50%.[60] Clinical picture resembles thalassemia Intermedia. There is increase in Hb F and HbS levels and prognosis is better than Thalassemia major and Sickle cell anemia.

b) HbE thalassemia- This results when patient inherit gene of beta thalassemia trait from one parent and HbE gene from other parent. Clinical and hematological features are variable. HPLC demonstrates HbA, HbF and Hb E in β+/E while no HbA in β0/E thalassemia. As such HbE trait may not be of clinical significance, but interaction of HbE and thalassemia produces variable phenotype.[61]

c) HbD thalassemia- There is interaction of Hb D and Beta thalassemia trait genes. HPLC reveals bands of HbA, HbF and HbD.

d) Delta Beta thalassemia- There is reduced and absent production of both delta and beta chains and increase in gamma chain synthesis. They are of 2 types. One type is associated with production of Hb Lepore and is usually 10-14%. HbF is 10-20%. Hb Lepore is characterized by hump on downward slope on HPLC. Other type (heterozygous) demonstrate increased HbF is 5-20% and HbA2 is normal or low. Cases of homozygous Delta beta thalassemia has HbF only and is >95%. MCV, MCH are low with high RBC count and normal RDW.

e) Hereditary persistence of fetal Hb- Increased levels of HbF in adults. In homozygous condition red cells only contain HbF. HPLC demonstrates near total absence of HbA with HbF being>96% in homozygotes. In heterozygotes HbF is 10-30%.

Sickle cell disorders and other Hemoglobinopathies

Cases of homozygous state have mainly HbS in their RBCs and is most severe. Heterozygous patients reveal presence of both HbA and HbS in their red cells and significant clinical feature are absent. Sickling syndrome also include double heterozygotes with one gene of HbS and other gene of beta thalassemia, D or E hemoglobin.

HbS is prevalent in Africa, Mediterranean countries and India. It has been observed that there is high prevalence of HbS in areas which are endemic for falciparum malaria suggesting HbS provides protection against falciparum malaria.

Structure of Hemoglobin S

The sickle mutation is caused by adenine to thymidine substitution in codon 6 of the beta globin gene. It results in substitution of valine in place of glutamic acid in the 6th position of beta chain.

In India HbS gene is mostly confined to tribes in Central-Eastern India where this disease is prevalent [62,63] and frequency ranges from 5-35%. WHO worldwide estimates about 20-25 million homozygous individuals for sickle cell disorders, 5-10 million are in India.[30] Sickle cell carrier who is virtually symptom free contains <50% of HbS in red cells. Presence of other Hb like HbF reduces while HbC and HbD increase the propensity of sickling. Sickle cells have altered rheological properties they do not alter their shape predisposing to stasis and vascular occlusion.

Genetics of Sickle cell disorders

1) **Sickle cell anemia-** Homozygous state SS one gene each from both parents, manifest early in life since Hb S >70% in red cells.
2) **Sickle cell trait-** Heterozygous state AS HbS from one parent and HbA from other parent. Amount of HbS is 25-40%
3) **Sickle cell disease-** Disease states in which at least one gene is HbS which may be in combination with normal or abnormal gene.

Laboratory Diagnosis

a) CBC- Hb-5-10gm% TLC is elevated and platelet count is elevated. Reticulocytosis- 3-10%.
b) Peripheral smear shows moderate to severe anisopoikilocytosis, normocytic normochromic to mildly hypochromic, sickle cells, target cells and ovalocytes
c) Bone marrow shows erythroid hyperplasia with normoblastic reaction.

Diagnostic tests

a) Sickling test- In homozygous sickle cell anemia diagnosis is by confirming the presence of HbS which can be demonstrated using reducing agent like 2% sodium metabisulphite or sodium dithionite.
b) HPLC- HbS has retention time of 4.30-4.50 min while that of HbD Punjab is 4.05-4.15min. HbSS/HbSA – In HbS abnormal Hb is HbS constituting 70-90%. HbF is 10-30% but Hb A is nil. This differentiates homozygous state from heterozygous state.

Sickling syndromes

1) **HbS Beta thalassemia-** Double heterozygote for S gene from one parent and Beta thalassemia from other parent
2) **HbSE disease-** In double heterozygote major Hb is HbS (60-70%) and minor is HbE
3) **HbSD Punjab disease-** Double heterozygous for S and D Punjab disorder and is associated with significant sickling. HbS is 25-50%, HbD is 40-50% with mild elevation of HbF. Diagnosis is possible on HPLC.

Other hemoglobinopathies

1) HbD Punjab- HbD trait (1%) seen in patients from northern part of country.[64] Also known as HbD Los Angeles, commonest hemoglobinopathy in India. In beta chain there is substitution at position 121. Retention time on HPLC is 4.05-4.15min. HbD Punjab and HbD Iran have same electrophorectic mobility but different retention times.

2) HbD Iran- Mild disease, On HPLC, retention time (3.62min) is in the region of HbE (3.72min) and if electrophoresis is not carried out can be diagnosed as HbE trait. On HPLC- HbD Iran >40% while HbE <30% and Hb Lepore (3.5min) is <20%. Hb D Iran shows broad based curve which does not touch the baseline.

3) HbE disorders- Substitution of glutamic acid by Lysine at position 26 of Beta chain. After HbS it is next prevalent abnormal Hb in the world. On HPLC, A2 and E elute at the same time. HbE trait is diagnosed by

the presence of high HbA2 (E+A2) approximately 30%.[65] Retention time of HbE is 3.69-3.79min. The patients having HbE trait and HbE-β thalassemia were mainly from North- east India where this abnormal HbE and variants are highly prevalent.[62,66,67]

Alpha chain variant hemoglobins

HbQ India- Rare hemoglobinopathy with prevalence of 0.4% in Indian subcontinent.[68] It was described by Vella et al [69] in 1958 in association with alpha-thalassemia in a Chinese patient. The first case was reported by Sukumaran.[70] Found predominantly in individuals from western and northern India and is more common in the Sindhi population.[71] Variant of alpha thalassemia, electrophorectic mobility is in the region HbS/HbD. On HPLC – the elution peak is in unknown window at 4.70-4. 90min and peak is sharp. Individuals inheriting HbQ India are clinically silent with normal or mildly deranged hematological parameters and are incidentally detected during population or family screening.[68,72,73]

HbJ Meerut- alpha variant, On HPLC a sharp peak is seen in P3 window with retention time of 1.6-1.85min. Levels upto 6% are normal and HbJ >12% is significant.

HPLC Instrument

Cation exchange High Performance Liquid Chromatography (HPLC) emerged as one of the best methods for screening and detection of various hemoglobinopathies with rapid and precise results.[10] The simplicity of the automated system with internal sample preparation, superior resolution, rapid assay time and accurate quantification of Hb fractions make it ideal for routine clinical laboratory.[74] It has origin in late 1970, came into practice into 1980. At present cation exchange HPLC is the method of choice for measuring HbA2, HbF and for detection of other Hb variants. Widely used machine is the Hemoglobin Testing system (Biorad D10). It is recommended for detection of β-thalassemia trait in population and necessary for genetic counselling to reduce the incidence and burden of thalassemia major in the society.[75,76]

In study conducted by Khera et.al in Maulana Azad medical college, New Delhi, a total of 110 cases were diagnosed as thalassemia and hemoglobinopathies of which 87 were thalassemic disorders and 23 were of hemoglobinopathies. Out of 87 cases of thalassemic disorders majority (62 cases) were thalassemia trait and out of 23 cases of hemoglobinopathy majority (8 cases) were HbD Punjab trait.[77]

In another study conducted by Iqbal et al, out of 518 samples, 111 samples showed abnormal types of Hb. There were 57 males and 54 female patients. The most common abnormal Hb disorder detected was Sickle cell trait followed by beta-thalassemia trait. The mean sickle cell Hb in Sickle cell trait was 33% and mean HbA2 in Beta-thalassemia trait was 5.2% respectively. The major studies on hemoglobinopathies and thalassemia confined only to few regions and revealed varying frequencies of Beta-thalassemia trait and other abnormal types of Hb.[78]

Mukhopadhyay et al in West Bengal screened a total of 10407 subjects and reported a prevalence of 5.6% for Beta-thalassemia trait and 5.0% carriers for HbE.[79] In yet another study conducted by Bhalodia et al reported a prevalence of 5.2% and 1.2% for Beta-thalassemia trait and sickle cell trait respectively.[80]

Singh et al studied 100 cases, of these, 51 cases showed abnormal Hb disorders and out of which 32 were females and 19 were males. The most common Hb disorder in their study was Beta-thalassemia trait.[81] which is in accordance with study conducted by Chandrashekhar V et al But more when compared with Rao S et al and Hosseini et al.[6,82,83]

Chopra et al conducted a study where most common hemoglobinopathy was Beta-thalassemia trait followed by sickle cell trait. The patients of Beta-thalassemia trait were from all parts of country and the patients of sickle cell trait and sickle cell disease were mostly from Central eastern India. Patients having HbE trait, HbE disease and HbE-β thalassemia were mainly from central coastal and North East India where abnormal HbE and variants are highly prevalent. HbD trait was seen in patients from Northern part of country.[84]

Pachori et al conducted a study for 5 years and found that overall hemoglobinopathies were more common in males than in females which was similar to study conducted by Dangi CBS et al (2013). However Beta-thalassemia trait and HbD-β thalassemia were diagnosed only in females and HbD trait was more common in females than males.[85]

Narang et al conducted a study for two years and found that out of 200 cases, 78 cases had one or other hemoglobinopathies. Most common among them were sickle thalassemia (56.4%) followed by beta-thalassemia trait (16.6%), sickle cell trait (15.3%), beta-thalassemia (10.2%) and sickle cell disease (1.3%). Several other studies have documented such type of frequency of hemoglobinopathy.[86]

In a study conducted by Harrison et al in a tertiary care teaching hospital in North India for a period of seven years for screening of Hb disorders, Of a total 7530 patients screened, 31 cases were diagnosed to have HbQ India and out of 31 cases, 25 cases had HbQ India trait and 6 cases were compound heterozygous for HbQ India and Beta-thalassemia trait.[87]

In a study conducted in northern India and Nepal in 2010, on 2600 patients, the prevalence of HbQ India was reported to be 0.2%. Panigrahi et al[88] in their study on 4500 patients in 2005 reported a prevalence of 0.4%.

In a study conducted by Goswami et al in North Bengal medical college and hospital in 2010, Out of 1872 cases 890 were abnormal cases and of which 54.5% were males and 45.5% were females. HbE trait was the most common hemoglobinopathy, followed by Homozygous HbE and Beta-thalassemia trait.[89]

In another study conducted for 15 years by Nadkarni et al 31075 patients were screened out of which 14423 individuals (46.4%) showed Hb defects. Heterozygosity for Beta thalassemia was seen in 27.4% and 7% show heterozygosity for sickle cell anemia. Homozygosity and compound heterozygosity for different Hb disorders ranges between 0.001 to 2.1%. Genotyping for beta thalassemia was carried out in 5615

beta thalassemia heterozygotes, out of 49 different mutations, 7 common Indian mutations account for 89.8% of molecular defects. IVS 1-5 (G>C) was the most common mutation (55.4%).[90]

In a one-year study, in Apollo Hospitals, Chennai, a tertiary care hospital in South India, conducted on 543 patients, the commonest disorder was β-thalassemia trait (37.9%) followed by HbE trait (23.2%), homozygous HbE (18.9%), HbS trait (5.3%), HbE β-thalassemia (4.6%), HbS β-thalassemia (2.5%), β-thalassemia major(2.3%), HbH(1.6%), homozygous HbS (1.4%), HbD trait(0.7). Of these 53.2% were males and 46.7% females. On comparing the thalassemic disorders (β-thalassemia trait, β-thalassemia major, and α-thalassemias), it was observed that the lowest average hemoglobin was seen in β-thalassemia major. The lowest average MCV was seen in β-thalassemia trait, whereas the lowest average MCH and MCHC in α-thalassemias. The average HbF was 88% in β-thalassemia major and less than one in α-thalassemias (HbH).[91]

MATERIALS AND METHODS

SOURCE OF DATA

The present 2.5 years (1 year retrospective and 1.5 years prospective) descriptive study was conducted in the Department of Pathology, Dayanand Medical College and Hospital, Ludhiana. All blood samples received in the immunopathology lab for High Performance Liquid Chromatography (HPLC) were analysed.

METHOD OF COLLECTION OF DATA

Inclusion criteria

All blood samples received in immunopathology lab for HPLC.

Exclusion criteria

None

Duration of Study

This study was conducted over a period of two and half years (1st January 2018 to 30th June 2020).

Study Design

This was a Prospective and Retrospective study

Methodology

Clinical history, Physical examination and investigations of all patients having abnormal HPLC chromatograms were recorded as per proforma. The cases with thalassemia or hemoglobinopathy were further investigated for family history and molecular testing.

Sample collection and analysis.

Peripheral blood sample in EDTA vial was collected. Routine complete blood counts were assessed using automated analyzers (Coulter LH 750 Analyzer/ Unicel DXH 800 Coulter cellular Analysis System/ Horiba ABX

Micros ES 60). The instrument works on the principle of Electrical impedance (Coulter principle). Cells are sized and counted by detecting and measuring changes in electrical resistance when a particle passes through a small aperture. Height of the pulse generated is proportional to cell volume (size) and the number of pulses indicates cell number (count). Hb, TLC, DLC, Red cell indices, mentzer's index and platelets were analyzed using automated hematology analyzer. Peripheral smear was studied after staining with Giemsa stain or other stains whenever required.

High Performance Liquid Chromatography was performed from the same sample (3 ml EDTA vial) using Biorad variant D-10 instrument.

Principle and procedure: A dual-piston, low pulsation High Performance Liquid Chromatography pump and a proportionating valve deliver the buffer solution to the analytical cartridge and detector. Whole blood samples undergo an automatic two-step dilution process and are introduced into the analytical flow path. Prediluted samples are identified based upon the use of a sample vial adapter in the sample rack, and the automatic dilution step is omitted. Pre diluted samples are aspirated directly and introduced into the analytical flow path. Between sample injections, the sample probe is rinsed with Wash/Diluent Solution to minimize sample carryover.

A Programmed phosphate ion gradient generated by mixing 2 buffers of increasing ionic strength delivers the sample to the analytical cartridge, where the different hemoglobins are separated based upon their ionic interactions with the cartridge material. The separated hemoglobins then pass through the photometer flow cell where changes in absorbance are measured at 415nm. A secondary filter at 690 nm corrects the effects caused by mixing buffers of different ionic strengths.

The software performs a reduction of raw data collected from each analysis that may include use of a calibration factor. A sample report and chromatogram where the different peaks are identified in defined

windows with relevant information like retention time, relative percentage and area.

STATISTICAL ANALYSIS

Data were described in terms of range; mean ±standard deviation (± SD), frequencies (number of cases) and relative frequencies (percentages) as appropriate. For comparing categorical data, Chi square ($\chi 2$) test was performed. A probability value (p value) less than 0.05 was considered statistically significant. All statistical calculations were done using (Statistical Package for the Social Science) SPSS 21version (SPSS Inc., Chicago, IL, USA) statistical program for Microsoft Windows.

OBSERVATIONS AND RESULTS

The present two and half year study (1st January, 2018 to 30th June 2020) to analyse the spectrum of hemoglobinopathies and thalassemia was conducted in the Department of Pathology, Dayanand Medical College and Hospital, Ludhiana. During this period a total of 3135 blood samples were analysed using Bio Rad D10 instrument for respective hemoglobin components (Hb A, A2, F,S,C, etc) using High Performance Liquid Chromatography (HPLC) studies. Clinical Profile, Hematological parameters and other investigations of all identified cases of thalassemia and hemoglobinopathies were done as per performa.

Of the total 3135 samples evaluated, 405 patients had either increased percentages of hemoglobins or had abnormal unknown peaks at various retention times. Remaining samples with normal HPLC were not analyzed further.

Age and Sex Distribution of all Cases with Abnormal HPLC Patterns

Of all the 405 cases, majority patients were in age group of 21 to 30 years (181 cases, 44.5%) followed by age group of 31 to 40 years (102 cases, 25.2%) (Table2, Fig.2) and had mean age of 28.5 years with youngest being one month old female child and eldest being 72 years male. The cases of abnormal hemoglobins included 137 males (33.8%) and 268 females (66.2%). Overall female preponderance was noted with M:F= 1:1.9 (Table3, Fig.3).

Table 2: Age wise distribution of cases

Age group (years)	No. of cases (n=405)	Percentage (%)
0-10	48	11.9
11-20	28	6.9
21-30	181	44.7
31-40	102	25.2
41-50	16	4.0
51-60	17	4.2
61-70	10	2.5
71-80	03	0.7

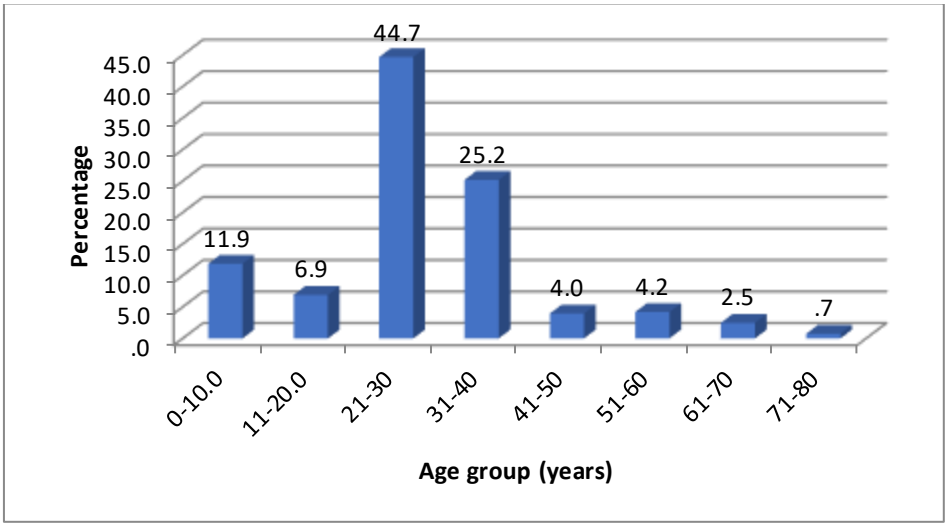

Fig. 2: Age wise distribution of cases

Table 3: Gender wise distribution of cases

Gender	No. of cases	Percentage (%)
Male	137	33.8
Female	268	66.2
Total	405	100

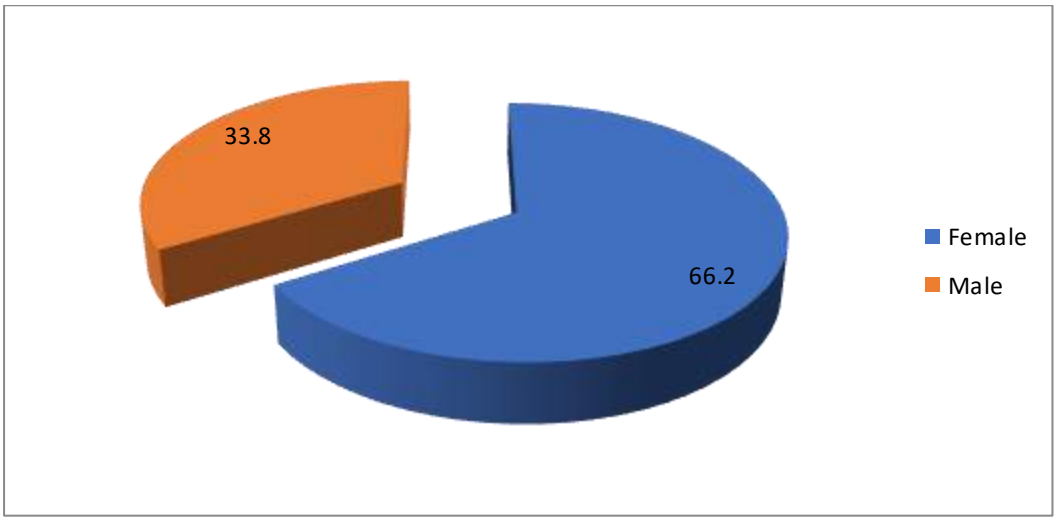

Fig 3: Gender wise distribution of cases

Distribution of cases according to Abnormal HPLC patterns

Of the 405 cases, majority were Beta thalassemia trait (63.0%), followed by HbD Punjab heterozygous (9.3%), Beta thalassemia homozygous (5.4%), Hb Q India heterozygous (1.5%), Compound heterozygous for Hb D Punjab and Beta thalassemia (1%). 2 cases were diagnosed each of Sickle cell heterozygous, Sickle cell homozygous, HbE heterozygous and HbD Iran heterozygous and compound heterozygous for HbQ and β thalassemia (0.5% each). Also one case each of HbJ Meerut, Compound heterozygous for HbS, HbE and HbJ with β thalassemia were diagnosed along with Compound heterozygous for HbS with delta β thalassemia. There were 65 cases (16.1%) which were diagnosed as Borderline (Table 4).

Table 4: Spectrum of thalassemia and hemoglobinopathies

HPLC Diagnosis	No of cases	Percent (%)
β-THALASSEMIA TRAIT	255	63.0%
HbD PUNJAB HETEROZYGOUS	38	9.3%
β-THALASSEMIA HOMOZYGOUS	22	5.4%
HbQ INDIA HETEROZYGOUS	06	1.5%
COMPOUND HETEROZYGOUS FOR HbD PUNJAB & β THALASSEMIA	04	1%
SICKLE CELL HETEROZYGOUS	02	0.5%
SICKLE CELL HOMOZYGOUS	02	0.5%
HbE HETEROZYGOUS	02	0.5%
HbD IRAN HETEROZYGOUS	02	0.5%
COMPOUND HETEROZYGOUS FOR HbQ & β THALASSEMIA	02	0.5%
COMPOUND HETEROZYGOUS FOR HbS & β THALASSEMIA	01	0.2%
COMPOUND HETEROZYGOUS FOR HbE & β THALASSEMIA	01	0.2%
HbJ MEERUT	01	0.2%
COMPOUND HETEROZYGOUS FOR HbJ & β THALASSEMIA	01	0.3%
COMPOUND HETEROZYGOUS FOR HbS & DELTA β THALASSEMIA	01	0.3%
BORDERLINE	65	16.1%
Total	405	100%

1) BETA THALASSEMIA

a) Beta thalassemia trait (BTT)

Of the total 405 cases majority (255/405, 63%) were diagnosed as Beta thalassemia trait. Majority were in the age group of 21 to 30 years with

youngest being 6 months old male and eldest being 72 years female and had slight female preponderance with M:F ratio 1:1.6.

Diagnosis

On high performance liquid chromatography HbA2 values more than 4% and less than equal to 10% were characterised as diagnostic of Beta thalassemia trait. In our study HbA2 ranged from 4.0% to 10.2% in these patients with a mean of 5.6%. The retention time for HbA2 ranged between 3.21 to 3.32 minutes. In males HbA2 ranged from 4.0% to 10.2% with a mean of 5.7% and in females HbA2 ranged from 4% to 7.7% with a mean of 5.5%. Of the 255 cases, majority cases had HbA2 ranging from 4% to 6.9% (96%), followed by 3.5% cases with HbA2 between 7% to 9% and 0.5% cases with HbA2>9%. HbF ranged from 0.2% to 16% with a mean of 1.57% Most of the cases had HbF<5% (94.5%), followed by 4.2% cases with HbF between 5% to 10% and 1.3% cases with HbF>10%.

HbA ranged from 63.6% to 95.9% with a mean of 86.2%. Majority cases had HbA ranged from 80% to 90% (46.2%), followed by 41% cases with normal range HbA (>90%) and 12.8% cases with HbA<80% (Table 6)

Clinical Profile

Pallor was noted in 138 patients (54.1%), Icterus in 8 patients (3.1%), 14 patients required blood transfusion (5.5%) and 3 had splenomegaly (1.1%)

Hematological parameters

Hb levels showed a wide range from 3.3 to 14.9 g/dl with a mean of 10.33 g/dl. Majority had anemia (79.9%) with mild, moderate and severe anemia in 14.2%, 56.2% and 9.5% respectively. On peripheral smear examination majority revealed microcytic hypochromic RBC picture with few target cells.

On evaluating other hematological parameter it was observed that RBC count ranged from 1.51 to 7.55x10^6/µL (Normal range 4.5 -5.5x10^6/µL)

with a mean of $5.17x10^6$/µL. Majority cases had normal range RBC count (43.0%), followed by 35.8% cases having high RBC count (>$5.5x10^6$/µL) and 21.2% cases had RBC count <$4.5x10^6$/µL. MCV ranged from 46 to 119 fL (Normal range 80-100fL) with a mean of 65.80fL. Majority cases had low MCV< 80 fl(95.2%) followed by 4.3% cases having normal range MCV and 0.5% cases had high MCV(>100 fL). Mentzer's Index was calculated as MCV (in fL) divided by RBC count (in millions/µl). Value <13 suggests thalassemia trait, >13 suggests Iron deficiency anemia. It ranged from 2.6 to 51.3 with a mean of 13.4. In males it ranged from 2.6 to 51.3 with a mean of 12.5 and in females it ranged from 8.1 to 35.6 with a mean of 14.0. Majority cases had Mentzer's index<13 (62.4%) and 37.6% cases had Mentzer's index>13.

MCH ranged from 11.8 to 39.9 pg (Normal range 27-32 pg) with a mean of 20.58 pg. Majority cases had low MCH < 27 pg (94.5%), followed by 4.7% cases having normal range MCH and 0.8% cases had MCH>32 pg. MCHC ranged from 19.9 to 34.3 g/dl (Normal range 32-37g/dl) with a mean of 30.62 g/dl Majority cases had MCHC< 32 g/d (91.0%) and 9.0% cases had MCHC>32 g/dl. RDW showed wide range from 13.6% to 35% (normal range 12-15%) with a mean of 17.63%. Majority had RDW between 15 to 20%(82.3%) followed by 12.2% cases having RDW>20 and 5.5% cases had normal range RDW**.** (Table 6)

Sensitivity and Specificity of HPLC instrument was found to be 98.8% and 37.1% respectively and accuracy was found to be 68.1% (Table 5)

Table 5: Comparison of HbA2 values with Mentzer's Index to calculate sensitivity, specificity and accuracy of HPLC.

HbA2	Mentzer's Index		Total	P- value
	<13	>13		
>4	159	100	259	
<4	2	59	61	
Total	161	159	320	0.001

Table 6: Different Hb fractions on HPLC and hematological parameters in β thalassemia trait cases

			No of cases	Percentage(%)
Different Hb fractions (%)	**HbF (%)**	**a) <5**	241	94.5
		b) 5-10	11	4.2
		c) >10	03	1.3
	HbA2 (%)	**a) 4-6.9**	245	96
		b) 7-9	09	3.5
		c) >9	01	0.5
	HbA (%)	**a) <80**	34	13.3
		b) 80-90	118	46.2
		c) >90	103	40.5
Hematological Parameters	**Hb (g/dl)**	**a) ≥ 12**	51	20.1
		b) 11-11.9	36	14.2
		c) 8-10.9	144	56.2
		d) <8	24	9.5
	RBC Count(10^6/µL) -> Normal range 4.5 -5.5x10^6/µL			
		a) <4.5	54	21.2
		b) 4.5-5.5	110	43.0
		c) >5.5	91	35.8
	MCV (fL) -> Normal range 80-100fL			
		a) <80	243	95.2
		b) 80-100	11	4.3
		c) >100	01	0.5
	MCH (pg) -> Normal range 27-32 pg			
		a) <27	241	94.5
		b) 27-32	12	4.7
		c) >32	02	0.8
	MCHC (g/dl)-> Normal range 32-37 g/dl			
		a) <32	232	91
		b) 32-37	23	9.0
	RDW (%) -> Normal range 12-15%			
		a) <15	14	5.5
		b) 15-20	210	82.3
		c) >20	31	12.2
	Mentzer's Index	**a) <13**	159	62.4
		b) >13	96	37.6

* Hb levels >12g/dl in females and >13g/dl in males are normal

Molecular Genetics

Molecular analysis was undertaken to identify the molecular mutations. Genotyping for β thalassemia was carried out in 28 β thalassemia trait cases. Nine different mutations were characterized. However three most common Indian mutations (IVS1-5 (G->C), Fr-8/9 (+G) and (IVS1-1 (G->T) accounted for 71.4% of molecular defects. IVS1-5 (G->C) was the common mutation encountered (28.6%). Few other rare mutations were also identified. Spectrum of mutations identified were shown in (Table 7).

Table 7: Spectrum of β thalassemia mutations identified in β thalassemia trait

Sr. no.	Mutation	No of cases	Percentage (%)
1	IVS1-5 (G->C)	08	28.6
2	Fr-8/9 (+G)	07	25.0
3	IVS1-1 (G->T)	05	17.8
4	Codon 5 (-CT)	02	7.1
5	619 bp deletion	02	7.1
6	Initiation Codon (ATG->ACG)	01	3.6
7	Fr-41/42 (-TTCT)	01	3.6
8	-88 (C->T)	01	3.6
9	Codon 8/9 (+G)	01	3.6

b) Beta Thalassemia Homozygous

Of the 405, 22 were diagnosed as beta thalassemia homozygous (5.4%).The age group ranged from one month to 60 years with male to female ratio of 1:1.

Diagnosis

All the patients whose sample on HPLC showed marked increase in HbF with or without increase in HbA2 and concomitant marked reduction in HbA were diagnosed as beta thalassemia homozygous.

HPLC findings

HbF ranged from 20.1% to 97% with a mean of 68.41%. Majority cases had increased HbF (HbF>80) (50.0%), followed by 31.8% cases with HbF between 40% to 80% and 18.2% cases with HbF<40%. HbA2 ranged from 0.6% to 8.2% with a mean of 2.90%. Majority cases had HbA2<4% (86.4%), followed by 9.1% cases with HbA2 between 4% to 7% and 4.5% cases with HbA2>7%. HbA ranged from 0.4% to 72% with a mean of 27.20%. Majority cases had HbA ranging from 0% to 50% (72.7%), followed by only 27.3% cases with Hb>50% (Table 8)

Clinical Profile

Pallor was noted in 16 patients (72.7%), Icterus in 14 patients (63.6%), 20 patients had splenomegaly (91%) of whom 11 required blood transfusion. Fatiguibility and Generalized weakness was present in almost all patients

Haematological parameters

Hb levels showed a wide range from 2.7 to 10.1 g/dl with a mean of 6.36 g/dl. All cases had anemia with moderate and severe anemia in 27.3% and 72.7% respectively. Peripheral blood smear showed marked anisopoikilocytosis, microcytosis, hypochromia, few tear drop cells and target cells along with nucleated RBC's. RBC count ranged from 1.01 to $4.6x10^6/\mu L$ (Normal range 4.5 -$5.5x10^6/\mu L$) with a mean of $2.60x10^6/\mu L$. Majority cases had low RBC count<$4.5x10^6/\mu L$(90.9%), followed by 9.1% cases with normal range RBC count and no case had RBC count>$5.5x10^6/\mu L$. MCV ranged from 62 to 94fL (Normal range 80-100fL) with a mean of 73.05 fL. Majority cases had low MCV< 80 fl(86.3%) followed by 13.7% cases with normal range MCV. Mentzer's Index was

calculated as MCV (in fL) divided by RBC count (in millions/µl). It ranged from 14.2 to 74.1 with a mean of 33.6. Majority cases had Mentzer's index>13 (95.5%) and only one case (4.5%) had Mentzer's index<13

MCH ranged from 19.9 to 32.9pg (Normal range 27-32 pg) with a mean of 24.97 pg. Majority cases had MCH < 27 pg (63.6%), followed by 31% cases having normal range MCH and one case (4.6%) had MCH>32 pg. MCHC ranged from 26 to 44 g/dl (Normal range 32-37g/dl) with a mean of 32.17g/dl. Majority cases had MCHC < 32 g/dl (59.1%) and 40.9% cases had MCHC>32 g/dl. RDW showed wide range from 19% to 37% (Normal range 12-15%) with a mean of 28.05%. Majority cases had RDW>20% (95.5%) followed by one case (4.5%) having RDW between 15 to 20%. No case had normal range RDW. (Table 8)

Table 8: Different Hb fractions on HPLC and hematological parameters in β thalassemia homozygous cases

			No of cases	Percentage (%)
Different Hb fractions (%)	**HbF (%)**	**a) <40**	04	18.2
		b) 40-80	07	31.8
		c) >80	11	50.0
	HbA2 (%)	**a) <4**	19	86.4
		b) 4-7	02	9.1
		c) >7	01	4.5
	HbA (%)	**a) 0-50**	16	72.7
		b) >50	06	27.3
Hematological Parameters	**Hb (g/dl)**	**a) ≥ 12**	0	0
		b) 11-11.9	0	0
		c) 8-10.9	06	27.3
		d) <8	16	72.7
	RBC Count (x10^6/µL) -> Normal range 4.5 -5.5x10^6/µL			
		a) <4.5	20	90.9
		b) 4.5-5.5	02	9.1
		c) >5.5	0	0
	MCV (fL) -> Normal range 80-100fL			
		a) <80	19	86.3
		b) 80-100	03	13.7
		c) >100	0	0
	MCH (pg) -> Normal range 27-32 pg			
		a) <27	14	63.6
		b) 27-32	07	31.8
		c) >32	01	4.6
	MCHC (g/dl)-> Normal range 32-37 g/dl			
		a) <32	13	59.1
		b) 32-37	09	40.9
	RDW (%)-> Normal range 12-15%			
		a) <15	0	0
		b) 15-20	01	4.5
		c) >20	21	95.5
	Mentzer's Index	**a) <13**	01	4.5
		b) >13	21	95.5

* Hb levels >12g/dl in females and >13g/dl in males are normal

Genetics

Molecular analysis was undertaken to identify the molecular mutations. Genotyping for β thalassemia was carried out in 14 β thalassemia homozygous cases. Five different mutations were characterized. However three most common Indian mutations (IVS1-5 (G->C), Fr-8/9 (+G) and (IVS1-1 (G->T) accounted for 78.6% of molecular defects. Fr-8/9 (+G) was the commonest mutation encountered (42.9%). Few other rare mutations were also identified. Spectrum of mutations identified were shown in (Table 9).

Table 9: Spectrum of β thalassemia mutations identified in β thalassemia homozygous cases

Sr. no.	Mutation	No of cases	Percentage (%)
1	Fr-8/9 (+G)	06	42.9
2	IVS1-1 (G->T)	03	21.4
3	IVS1-5 (G->C)	02	14.3
4	Fr-41/42 (-TTCT)	02	14.3
5	Codon 5 (-CT)	01	7.1

2) HbD Punjab Heterozygous

Out of 405 cases, 38 cases were HbD Punjab heterozygous (9.3%). Age group ranged from one year to 54 years. Majority cases were in the age group of 31 to 40 years. Female preponderance noted with male to female ratio of 1:5.33.

Diagnosis

HbD ranged from 11% to 40.4% with a mean of 33.82% and retention time ranged from 3.83 to 3.89 minutes. Majority cases had HbD ≥30% (86.8%). 5 cases had HbD<30% (13.2%). HbF ranged from 0% to 1.6%

with a mean of 0.67%. Majority cases had HbF<1% (94.7%), Only two cases (5.3%) had HbF≥1%. HbA2 ranged from 1.7% to 3.5% with a mean of 2.72%. Majority cases had HbA2>2.5% (63.2%) and 36.8% cases had HbA2≤2.5%. HbA ranged from 49.8% to 85.3% with a mean of 60.51%. Majority cases had Hb>50 (97.4%) and only one case (2.6%) had HbA≤50% (Table 10).

Clinical Profile

Pallor was noted in 14 patients (36.8%), Icterus in only one patient (2.6%) and three patients required blood transfusion (7.9%).

Hematological Parameters

Hb levels showed a wide range from 3.9 to 16.3 g/dl with a mean of 10.99 g/dl. Half cases had anemia (49.9%) with mild, moderate and severe anemia in 10.5%, 26.3% and 13.1% respectively. Half of cases had normal range Hb (≥12g/dl). Peripheral blood smear showed microcytic hypochromic RBC's and few target cells. RBC count ranged from 1.38 to 6.07x10^6/µL (Normal range 4.5 -5.5x10^6/µL) with a mean of 4.09x10^6/µL. Majority cases had RBC count less than 4.5x10^6/µL (65.7%). Two cases had normal range RBC count (28.9%). MCV ranged from 52.1 to 112 fL (normal range 80-100fL) with a mean of 83.33 fL. Majority cases had normal range MCV. (63.1%). 11 cases had low MCV<80 fL (28.9%) and three cases had MCV>100 fL(8.0%). Mentzer's Index ranged from 10.7 to 75.1 with a mean of 23.20. Majority cases had Mentzer's index>13 (97.4%) and only one case (2.6%) had Mentzer's index<13

MCH ranged from 13 to 36.7 pg (Normal range 27-32 pg) with a mean of 27.11 pg. Majority cases had normal range MCH (55.3%). 14 cases had MCH<27pg (36.8%) and 3 cases had MCH>32 pg (7.9%). MCHC ranged 24.6 to 34.9 g/dl (Normal range 32-37 g/dl) with a mean of 32.33 g/dl. Majority cases had normal range MCHC (71.1%) and 11 cases had MCHC<32 g/dl (28.9%). RDW ranged from 12.9% to 35.7% (Normal range 12-15%) with a mean of 17.76%. Majority cases had mild degree

of anisocytosis (42.1%). 39.5% cases had normal range RDW and 18.4% cases had moderate degree of anisocytosis. (Table 10)

Table 10: Different Hb fractions on HPLC and hematological parameters in HbD Punjab heterozygous cases.

			No of cases	Percentage(%)
Different Hb fractions (%)	**HbD (%)**	**a) <30**	05	13.2
		b) ≥30	33	86.8
	HbF (%)	**a) <1.0**	36	94.7
		b) ≥1.0	02	5.3
	HbA2 (%)	**a) ≤2.5**	14	36.8
		b)>2.5	24	63.2
	HbA (%)	**a) ≤50**	01	2.6
		b) >50	37	97.4
Hematological Parameters	**Hb (g/dl)**	**a) ≥ 12**	19	50.1
		b) 11-11.9	04	10.5
		c) 8-10.9	10	26.3
		d) <8	05	13.1
	RBC Count(x10^6/µL) -> Normal range 4.5 -5.5x10^6/µL			
		a) <4.5	25	65.7
		b) 4.5-5.5	02	28.9
		c) >5.5	0	5.4
	MCV (fL) -> Normal range 80-100fL			
		a) <80	11	28.9
		b) 80-100	24	63.1
		c) >100	03	8.0
	MCH (pg) -> Normal range 27-32 pg			
		a) <27	14	36.8
		b) 27-32	21	55.3
		c) >32	03	7.9
	MCHC (g/dl)-> Normal range 32-37 g/dl			
		a) <32	11	28.9
		b) 32-37	27	71.1
	RDW (%)-> Normal range 12-15%			
		a) <15	15	39.5
		b) 15-20	16	42.1
		c) >20	07	18.4
	Mentzer's Index	**a) <13**	01	2.6
		b) >13	37	97.4

* Hb levels >12g/dl in females and >13g/dl in males are normal

3) **HbQ India heterozygous**

Six cases were diagnosed as HbQ India heterozygous (1.5%). The age group ranged from 24 to 53 years. Female preponderance was noted with male to female ratio of 1:5.

Diagnosis

Abnormal HbQ ranged from 15.3% to 20.3% with a mean of 16.7% and a retention time ranged from 4.35 to 4.45 minutes. Majority cases had HbQ<20% (83.3%) and one case had HbQ≥20% (16.7%). HbF ranged from 0% to 0.8% with a mean of 0.25%. All cases had HbF<1%. HbA2 ranged from 2.1% to 3.7% with a mean of 2.66%. Majority cases had HbA2≤3% (83.3%). Only one case had HbA2>3% (16.7%). HbA ranged from 67.9% to 81.9% with a mean of 75.28%. Half cases had HbA≤75% (50%). Half cases had HbA>75% (50%). (Table 11)

Hematological Parameters

Hb levels ranged from 8.3 to 12.9 g/dl with a mean of 11.55 g/dl. Majority cases had normal range (≥12 g/dl) Hb (66.6%). One case each had mild and moderate anemia respectively. Peripheral blood smear showed microcytic hypochromic RBC picture.

RBC count ranged from 3.98 to 4.81x10^6/µL (Normal range 4.5 - 5.5x10^6/µL) with a mean of 4.23x10^6/µL. Majority cases had RBC count<4.5x106/µL (83.3%). One case had normal range RBC count (16.7%). MCV ranged from 68.6 to 93.5 fL (Normal range 80-100fL) with a mean of 81.73 fL. Majority cases had normal range MCV (66.7%). Two cases had low MCV<80fL (33.3%). Mentzer's Index ranged from 14.6 to 23.4 with a mean of 19.4. All cases had Mentzer's index>13 (100%).

MCH ranged from 20.8 to 30.6pg (Normal range 27-32 pg) with a mean of 26.55 pg. Half cases had MCH<27 pg (50%) and half cases had normal range MCH (50%). MCHC ranged 30.3 to 33.7 g/dl (Normal range 32-37 g/dl) with a mean of 32.4 g/dl. Majority cases had normal range MCHC (83.3%) and one case had MCHC<32 g/dl (16.7%). RDW ranged from 12 % to18.7 % (Normal range 12-15%) with a mean of

15.46%. Majority cases had mild degree of anisocytosis (66.7%) and two cases had normal range RDW (33.3%). (Table 11)

Table 11: Different Hb fractions on HPLC and hematological parameters in HbQ India heterozygous cases.

			No of cases	Percentage(%)
Different Hb fractions (%)	**HbQ (%)**	**a) <20**	05	83.3
		b) ≥20	01	16.7
	HbF (%)	**a) <1.0**	06	100
		b) ≥1.0	0	0
	HbA2 (%)	**a) ≤3**	05	83.3
		b)>3	01	16.7
	HbA (%)	**a) ≤75**	03	50.0
		b) >75	03	50.0
Hematological Parameters	**Hb (g/dl)**	**a) ≥ 12**	04	66.6
		b) 11-11.9	01	16.7
		c) 8-10.9	01	16.7
		d) <8	0	0
	RBC Count (x10^6/µL) -> Normal range 4.5 - 5.5x10^6/µL			
		a) <4.5	05	83.3
		b) 4.5-5.5	01	16.7
		c) >5.5	0	0
	MCV (fl)-> Normal range 80-100fL			
		a) <80	02	33.3
		b) 80-100	04	66.7
		c) >100	0	0
	MCH (pg) -> Normal range 27-32 pg			
		a) <27	03	50
		b) 27-32	03	50
		c) >32	0	0
	MCHC (g/dl) -> Normal range 32-37 g/dl			
		a) <32	01	16.7
		b) 32-37	05	83.3
	RDW (%)-> Normal range 12-15%			
		a) <15	02	33.3
		b) 15-20	04	66.7
		c) >20	0	0
	Mentzer's Index	**a) <13**	0	0
		b) >13	06	100

* Hb levels >12g/dl in females and >13g/dl in males are normal

4) Compound Heterozygous for HbD Punjab & β Thalassemia

Four cases were diagnosed as Compound Heterozygous for HbD Punjab & β Thalassemia (1%). The age group ranged from 28 to 72 years. M:F ratio was 1:1.

Diagnosis

HbD ranged from 37.1% to 83.7% with a mean of 70.1% and retention time ranged from 3.83 to 3.89 minutes. Majority cases had HbD≥70% (75%). Only one case had HbD<70% (25%). HbF ranged from 0% to 4.2% with a mean of 2.2%. Half cases had HbF<1% (50%) and half cases had HbF≥1% (50%). HbA2 ranged from 4% to 6.1% with a mean of 4.7%. All cases had HbA2≥4% (100%). HbA ranged from 6% to 58.2% with a mean of 21.5%. Majority cases had HbA≤50% (75%). Only one case had HbA>50% (25%). (Table 12)

Hematological Parameters

Hb levels ranged from 9.5 to 15.9 g/dl with a mean value of 11.82 g/dl. Majority cases had anemia (75%), with half cases having moderate degree of anemia (50%) and one case had mild anemia (25%). One case had normal range (≥12 g/dl) Hb (25%). RBC count ranged from 4.41 to 6.52x10^6/µL (Normal range 4.5 -5.5x10^6/µL) with a mean of 5.36x10^6/µL. Majority cases had RBC count >5.5x10^6/µL (50%). One case had RBC count <4.5x10^6/µL (25%) and one case had normal range RBC count (25%). MCV ranged from 54.8 to 83.8 fL (Normal range 80-100fL) with a mean of 69.8 fL. Majority cases had low MCV< 80 fL (75%). One case had normal range MCV (25%). Mentzer's Index ranged from 8.4 to 17.3 with a mean of 13.4. Majority cases had Mentzer's index>13 (75%). One case had Mentzer's index<13 (25%).

MCH ranged from 17.2 to 28.3 pg (Normal range 27-32 pg) with a mean of 22.27pg. Majority cases had MCH< 27 pg (75%). One case had normal range MCH (25%). MCHC ranged from 30 to 33.7 g/dl (Normal range 32-37 g/dl) with a mean of 31.75 g/dl. Majority cases had MCH<

32 g/dl (75%). One case had normal range MCHC (25%). RDW ranged from 14.6% to 24.1% (Normal range 12-15%) with a mean of 18.1%. Majority cases had mild degree of anisocytosis (50%). One case had moderate degree of anisocytosis (25%) and one case had normal range RDW (25%). (Table 12)

Table 12: Different Hb fractions on HPLC and hematological parameters in Compound heterozygous for HbD Punjab and β Thalassemia cases.

			No of cases	Percentage(%)
Different Hb fractions (%)	**HbD (%)**	**a) <70**	01	25
		b) ≥70	03	75
	HbF (%)	**a) <1.0**	02	50
		b) ≥1.0	02	50
	HbA2 (%)	**a) <4**	0	0
		b)≥4	04	100
	HbA (%)	**a) ≤50**	03	75
		b) >50	01	25
Hematological Parameters	**Hb (g/dl)**	**a) ≥ 12**	01	25
		b) 11-11.9	01	25
		c) 8-10.9	02	50
		d) <8	0	0
	RBC Count (x10^6/µL) -> Normal range 4.5 - 5.5x10^6/µL			
		a) <4.5	01	25
		b) 4.5-5.5	01	25
		c) >5.5	02	50
	MCV (fL) -> Normal range 80-100fL			
		a) <80	03	75
		b) 80-100	01	25
		c) >100	0	0
	MCH (pg) -> Normal range 27-32 pg			
		a) <27	03	75
		b) 27-32	01	25
		c) >32	0	0
	MCHC (g/dl)-> Normal range 32-37 g/dl			
		a) <32	03	75
		b) 32-37	01	25
	RDW (%)-> Normal range 12-15%			
		a) <15	01	25
		b) 15-20	02	50
		c) >20	01	25
	Mentzer's Index	**a) <13**	01	25
		b) >13	03	75

* Hb levels >12g/dl in females and >13g/dl in males are normal

Genetics

Molecular was done in one patient in which IVS1-5 (G->T) mutation was identified.

5) Sickle Cell Heterozygous

Two cases were diagnosed as sickle cell heterozygous (0.5%). Both were female.

Diagnosis

One case had HbF-0.8%, HbA - 58.6, HbA2- 3.2% and HbS-29.9% at a retention time of 4.14 minutes

Other case had HbF-1.6%, HbA – 64.2%, HbA2- 3.1% and HbS-31.1% at a retention time of 4.13 minutes.

Hematological Parameters

Peripheral smear shows mild anisopoikilocytosis with normocytic normochromic RBC's and few sickle cells.

One case had Hb value of 9.1 g/dl, other case had 10.9 g/dl. Both cases had anemia of moderate degree. RBC count $3.82x10^6$/µL.MCV, MCH, MCHC and RDW value of 87fL, 28.5pg, 32.7g/dl, 14.1% respectively.

Other case had RBC count $4.52x10^6$/µL.MCV, MCH, MCHC and RDW value of 68fL, 20.2pg, 29.8g/dl and 17.7% respectively

6) Sickle Cell Homozygous

Two cases were diagnosed as sickle cell homozygous (0.5%). Male to female ratio were 1:1

Diagnosis

One case had HbF-17.1%, HbA – 8.4%, HbA2- 3.5% and HbS-71% at a retention time of 4.11 minutes.

Other case had HbF-24.5%, HbA – 5.3%, HbA2- 1.6% and HbS-66% at a retention time of 4.12 minutes.

Hematological Parameters

Peripheral smear showed moderate to severe degree of anisopoikilocytosis with mild hypochromia, sickle cells and few target cells.

One case had Hb value of 11.9 g/dl, other case had 8.7 g/dl. One case had mild anemia and other case had moderate anemia. RBC count $4.31x10^6$/µL.MCV, MCH, MCHC and RDW value of 84fL, 27.5pg, 32.7g/dl, 16.6% respectively.

Other case had RBC count $2.87x10^6$/µL. MCV, MCH, MCHC and RDW value of 93fL, 30.1pg, 32.3g/dl and 16.8% respectively

7) HbE Heterozygous

Two cases were diagnosed as HbE heterozygous (0.5%). Male to Female ratio of 1:1 was noted.

Diagnosis

One case had HbF-0%, HbA – 75.5%, Unknown peak - 24.5% with retention time of 2.70 minutes.

Other case had HbF-0.7%, HbA – 66.5% and HbA2- 32.8% at a retention time of 3.13 minute.

Hematological Parameters

Peripheral smear showed microcytosis and few target cells.

One case had Hb value of 10.9 g/dl, other case had Hb levels of 13.7 g/dl. One case had RBC count $4.32x10^6$/µL. MCV, MCH, MCHC and RDW value of 78.7fL, 25.1pg, 32g/dl, 25.3% respectively. Other case had RBC count $5.32x10^6$/µL. MCV, MCH, MCHC and RDW value of 77.2fL, 25.8pg, 33.4g/dl and 16.6% respectively.

8) HbD Iran heterozygous

Two cases were diagnosed (0.5%). Both were female.

Diagnosis

One case had HbF-0%, HbA – 62.1% and HbA2- 40.9% at a retention time of 2.80 minutes. Other case had HbF-0.8%, HbA – 53.6% and HbA2- 44.5% at a retention time of 2.88 minutes.

Hematological Parameters

One case had Hb value of 9.5 g/dl, Other case had Hb levels of 11.3 g/dl. One case had RBC count $4.95x10^6$/µL.MCV, MCH, MCHC and RDW value of 66fL, 19.3pg, 29.3g/dl and 24.4% respectively. Other case had RBC count $4.07x10^6$/µL. MCV, MCH, MCHC and RDW value of 87fL, 27.8pg, 32.1g/dl and 15.5% respectively.

9) Compound heterozygous for HbQ India and Beta thalassemia

Two cases were diagnosed (0.5%). Male to female ratio of 1:1.

Diagnosis

One case had HbF-2.9%, HbA – 82.6%, HbA2- 6.7 and HbQ-7.8% at a retention time of 4.37 minutes. Other case had HbF-3.3%, HbA – 81.7% and HbA2- 5.5% and HbQ- 8.2% at a retention time of 4.35 minutes.

Hematological Parameters

One case had Hb value of 10.0 g/dl, Other case had Hb levels of 12 g/dl. One case had RBC count 4.88x106/µL.MCV, MCH, MCHC and RDW value of 64.5fL, 20.6pg, 31.9g/dl and 15.9% respectively. Other case had RBC count $5.49x10^6$/µL. MCV, MCH, MCHC and RDW value of 71.1fL, 21.9pg, 38.8g/dl and 22.4% respectively.

Genetics

Molecular was done in one patient in which 619 bp mutation was identified.

10) Compound heterozygous for HbS and Beta Thalassemia

Only one case was diagnosed and was male

Diagnosis

HbF value was 35.4%, HbA was 18.3%, HbA2 was 4% and HbS was 34.9% at a retention time of 4.16 minutes.

Hematological Parameters

One case was diagnosed which was male. Hb levels were 2.4g/dl and had severe degree of anemia. RBC count was $0.93x10^6$/µL. MCV, MCH and MCHC was 87.1fL, 26pg and 29.9g/dl respectively. RDW was 24.7%.

11) Compound heterozygous for HbE and Beta Thalassemia

One case was diagnosed which was female

Diagnosis

HbF value was 31.7%, HbA was 7.2%, HbA2 was 61.1% at a retention time of 3.02 minutes.

Hematological Parameters

Hb levels were 4.5g/dl and had severe degree of anemia. RBC count was $2.54x10^6$/µL. MCV, MCH and MCHC was 62fL, 17.7pg and 28.5g/dl respectively. RDW was 34.8% indicating severe degree of anisocytosis.

12) HbJ Meerut

One case was diagnosed and was female.

Diagnosis

HbA was 89.3%, HbA2 was 3.3% and P3 peak was 7.4% at a retention time of 1.47 minutes.

Hematological Parameters

Hb levels were 8.8g/dl and had moderate degree of anemia. RBC count was $3.41x10^6$/µL. MCV, MCH and MCHC was 77.3 fL, 25.7 pg and 33.2 g/dl respectively. RDW was 15.8%.

13) Compound heterozygous for HbJ and Beta thalassemia

One case was diagnosed which was female

Diagnosis

HbF was 2.1% HbA was 70.1%, HbA2 was 4.4% and P3 peak was 23.4% at a retention time of 1.57 minutes.

Hematological Parameters

Hb levels were 10.0 g/dl and had moderate degree of anemia. RBC count was $5.12x10^6/\mu L$. MCV, MCH and MCHC was 91 fL, 28.3 pg and 31 g/dl respectively. RDW was 31.2%.

14) Compound heterozygous for HbS and Delta Beta thalassemia.

One case was diagnosed which was male.

Diagnosis

HbF was 29.1%, HbA was 2.2%, HbA2 was 3.2% and HbS was 50.6 at a retention time of 4.11 minutes.

Hematological Parameters

Hb levels were 3.3 g/dl and had severe degree of anemia. Peripheral smear showed microcytic hypochromic RBC picture with few target cells.

RBC count was $1.17x10^6/\mu L$. MCV, MCH and MCHC was 63 fL, 19.5 pg and 31 g/dl respectively. RDW was 16.6%.

15) Borderline

Out of 405 cases, 65 were Borderline (16.1%). The age group ranged from 8 to 42 years. Majority cases were in the age group of 21-30 years. Majority were females with male to female ratio of 1:4.

Diagnosis

In our study HbA2 ranged from 3.6% to 4.2% with a mean of 3.75%. Majority cases had HbA2<4% (93.8%). Only four cases had HbA2≥4% (6.2%). HbF ranged from 0% to 6% with a mean of 0.55%. Majority cases had HbF<1% (95.4%) and HbF≥1% (4.6%). HbA ranged from 82.6% to 96.4% with a mean of 91.0%. Majority cases had HbA≥90% (57%). 43% cases had HbA<90%. (Table 13)

Hematological Parameters

Hb levels showed a wide range from 6.2 to 16.3 g/dl with a mean of 12.16 g/dl. Majority cases had normal range Hb (66.1%). 15.4% cases had moderate anemia followed by 12.3% cases which had mild anemia and only 6.2% cases had severe anemia. RBC count ranged from 1.77 to $5.49x10^6/\mu L$ (Normal range 4.5 -$5.5x10^6/\mu L$) with a mean of $4.26x10^6/\mu L$. Majority cases had RBC count less than $4.5x10^6/\mu L$ (53.8%), followed by 46.2% cases which had normal range RBC count. MCV ranged from 65.6 to 120.2 fL (Normal range 80-100fl) with a mean of 88.4 fL. Majority cases had normal range MCV (86.2%), followed by 10.8% cases which had MCV<80 fL and only 2 cases had MCV>100 (3.0%). MCH ranged from 20 to 39.6 pg (Normal range 27-32 pg) with a mean of 29.1 pg. Majority cases had normal range MCH (75.4%), followed by 16.9% cases which had MCH<27 pg and 7.7% cases had MCH >32 pg. MCHC ranged from 28.6 to 34.1 g/dl (Normal range 32-37 g/dl) with a mean of 32.5 g/dl. Majority cases had normal range MCHC (78.5%) and 21.5% cases had MCHC<32 g/dl. RDW ranged from 11.6% to 31.1% (Normal range 12-15%) with a mean of 15.0%. Majority of cases had normal range RDW (57.0%), followed by 41.5% cases which had mild degree of anisocytosis and only one case had moderate degree of anisocytosis (1.5%).Mentzer's Index ranged from 12.7 to 67.9 with a mean of 22.0. Majority cases had Mentzer's index>13 (97%). Only two cases had Mentzer's index<13 (3.0%) (Table 13). There were 3 cases in which inspite of HbA2≥4 there was raised HbF and are categorized under

borderline in which molecular genetics were done in 2 cases where they were for positive for IVS1-1 and Fr8/9 mutations respectively

Table 13: Different Hb fractions on HPLC and hematological parameters in Borderline cases

			No of cases	Percentage (%)
Different Hb fractions (%)	**HbA2 (%)**	**a) <4**	61	93.8
		b) ≥4	04	6.2
	HbF (%)	**a)<1**	62	95.4
		b) ≥1	03	4.6
	HbA (%)	**a) <90**	28	43.0
		b) ≥90	37	57.0
Hematological Parameters	**Hb (g/dl)**	**a) ≥ 12**	43	66.1
		b) 11-11.9	08	12.3
		c) 8-10.9	10	15.4
		d) <8	04	6.2
	RBC Count (x10^6/µL) -> Normal range 4.5 -5.5x10^6/µL			
		a) <4.5	35	53.8
		b) 4.5-5.5	30	46.2
		c) >5.5	0	0
	MCV (fL) -> Normal range 80-100fL			
		a) <80	07	10.8
		b) 80-100	56	86.2
		c) >100	02	3.0
	MCH (pg) -> Normal range 27-32 pg			
		a) <27	11	16.9
		b) 27-32	49	75.4
		c) >32	05	7.7
	MCHC (g/dl)-> Normal range 32-37 g/dl			
		a) <32	14	21.5
		b) 32-37	51	78.5
	RDW (%)-> Normal range 12-15%			
		a) <15	37	57.0
		b) 15-20	27	41.5
		c) >20	01	1.5
	Mentzer's Index	**a) <13**	02	3.0
		b) >13	63	97.0

* Hb levels >12g/dl in females and >13g/dl in males are normal

Table 14: Hematological parameters of abnormal HPLC patterns

HPLC Diagnosis	Hb(g/dl)	MCV(fL)	MCH (pg)	MCHC(g/dl)	RBC(10^6/µL)	RDW (%)
β-THALASSEMIA HETEROZYGOUS	10.3±1.8	65.7±8.0	20.7±4.5	30.6±1.4	5.2±5.1	17.6±3.2
β-THALASSEMIA HOMOZYGOUS	6.3±2.2	73.1±8.3	24.9±3.7	32.2±3.9	2.6±1.0	28.0±4.9
SICKLE CELL HETEROZYGOUS	10.0±1.3	77.5±13.4	24.4±5.9	31.2±2.0	4.2±0.5	15.9±2.5
SICKLE CELL HOMOZYGOUS	10.3±2.3	88.5±6.4	29.7±4.5	32.5±0.3	3.6±1.0	17.2±1.0
COMPOUND HETEROZYGOUS FOR HbS & β THALASSEMIA	2.41	87.1	26.0	29.9	0.9	24.7
HbE HETEROZYGOUS	12.3±2.0	77.9±1.0	25.4±0.5	32.7±0.9	4.8±0.7	20.9±6.2
COMPOUND HETEROZYGOUS FOR HbE& β THALASSEMIA	4.50	62.0	17.7	28.5	2.5	34.8
HbD PUNJAB HETEROZYGOUS	10.9±3.0	83.3±12.6	27.1±5.1	32.3±1.9	4.1±1.0	17.8±5.9

COMPOUND HETEROZYGOUS FOR HbD & β THALASSEMIA	11.8±2.8	69.8±12.9	22.3±5.0	31.7±1.5	5.4±0.9	18.1±4.2
HbQ INDIA HETEROZYGOUS	11.5±1.7	81.7±10.4	26.5±3.9	32.4±1.1	4.2±0.3	15.5±2.3
COMPOUND HETEROZYGOUS FOR HbQ INDIA & β THALASSEMIA	11.0±1.4	67.8±4.7	21.3±0.9	35.3±4.9	5.2±0.4	19.1±4.6
HbD IRAN HETEROZYGOUS	10.4±1.3	76.5±14.8	23.5±6.0	30.7±1.9	4.5±0.2	19.9±6.3
HbJ MEERUT	8.80	77.3	25.7	33.2	3.4	15.8
COMPOUND HETEROZYGOUS FOR HbJ & β THALASSEMIA	10.0	63.0	19.5	31.0	5.1	16.6
COMPOUND HETEROZYGOUS FOR HbS & DELTA β THALASSEMIA	3.3	91.0	28.3	31.0	1.2	31.2
BORDERLINE	12.2±2.1	88.5±9.2	29.1±8.7	32.5±1.0	4.3±0.7	15.6±4.7

All values were in mean±SD

Table 15: Different Hb fractions of abnormal HPLC patterns

HPLC Diagnosis	HbF (%)	HbA (%)	HbA2 (%)	Variant Hb (%)
β-THAL TRAIT	1.6±1.9	86.2±6.4	5.6±0.8	
β-THAL HOMOZYGOUS	68.4±26.6	27.2±25.9	2.9±1.5	
SICKLE CELL HETEROZYGOUS	1.2±0.5	61.4±3.9	3.1±0.0	30.5±0.8
SICKLE CELL HOMOZYGOUS	20.8±5.2	6.8±2.1	2.5±1.3	68.5±3.5
COMPOUND HETEROZYGOUS FOR HbS & β THAL	35.4	18.3	4.0	34.9
HbE HETEROZYGOUS	0.7	71.0±6.4	32.8	
COMPOUND HETEROZYGOUS FOR HbE & β THAL	31.7	7.2	61.1	
HbD PUNJAB HETEROZYGOUS	0.8	60.5±9.3	2.7±0.4	33.8±6.4
COMPOUND HETEROZYGOUS FOR HbD & β THAL	2.9	21.5±24.8	4.7±0.9	70.1±22.1
HbQ INDIA HETEROZYGOUS	0.7±0.3	75.3±5.5	2.7±0.6	16.7
COMPOUND HETEROZYGOUS FOR HbQ INDIA & β THAL	3.1±0.3	82.2±0.6	6.1±0.8	8.7
HbD IRAN HETEROZYGOUS	0.8	57.9±6.0	42.7±2.5	
HbJ MEERUT	.	89.3	3.3	7.4
COMPOUND HETEROZYGOUS FOR HbJ& β THAL TRAIT	2.1	70.1	4.4	23.4
COMPOUND HETEROZYGOUS FOR HbS& DELTA β THAL OR HPFH	29.1	2.2	3.2	50.6
BORDERLINE	0.6	91.0±5.4	3.7±0.1	

All values were in mean±SD

DISCUSSION

The present study was conducted in the Department of Pathology, Dayanand Medical College and Hospital, Ludhiana, for a period of two and half years on abnormal HPLC patterns. During this period a total of 3135 blood samples were analysed using Bio Rad D10 instrument for respective hemoglobin components (Hb A, A2, F ,S,C, etc) using high performance liquid chromatography (HPLC) studies.

Of the total 3135 samples evaluated, 405 patients had either increased percentages of hemoglobins or had abnormal unknown peaks at various retention times. Remaining samples with normal HPLC were not analyzed further.

Of the 405 cases included in the study majority were in the age group of 21 to 30 years (44.5%), followed by age group of 31-40 years (25.2%) which is in concordance with the study conducted by Singh et al.[81] Overall the age range of the cases included in this study group (1 month to 72 years) was similar to the studies conducted by Iqbal et al[78] and Shrivastav et al.[92] The mean age of cases was 28.5 years which is similar to studies conducted by Mondal et al[93] which had a mean age of 27.3 years. However, Goswami et al[89] and Shankar et al[94] had different findings where majority of cases were in the age group of 0 to 20 years and 0 to 15 years respectively.

In the present study abnormal hemoglobins were seen in 33.8% males and 66.2% females. Female preponderance has been reported which is in concordance with the study conducted by Mondal et al,[93] Singh et al[81] and Goswami et al.[89] It is in contrast to study conducted by Shankar et al[94] and Chopra et al[84] where male preponderance was noted. Narang et al study[86] found almost equal incidence in both sexes. The male to female ratio was 1:1.9 which is similar to studies conducted by Singh et al[81] and Mondal et al[93] where male to female ratio was 1:1.7 and 1:2.7 respectively. (Table 16)

The present findings of female preponderance and relatively younger age group being predominant could be because of HPLC being part of routine antenatal check-up in females of reproductive age when they are planning their families.

Table 16: Comparison of age and sex distribution with other studies

Authors	Age range (years)	M:F ratio
Mondal et al [93]	7 months to 69 years	1:2.65
Singh et al [81]	2 months to 40 years	1:1.68
Iqbal et al [78]	3 months to 74 years	1:1
Biswas et al [96]	1 year to 84 years	1.26:1
Sarvaiya et al [95]	28 days to 74 years	1.22:1
Present study	1 month to 72 years	1:1.9

In the present study, out of the 405 cases majority were Beta thalassemia trait (63%), followed by Hb D Punjab heterozygous (9.3%), Beta thalassemia homozygous (5.4%), Hb Q India heterozygous (1.5%), Compound heterozygous for HbD Punjab and Beta thalassemia (1%). Two case each of Sickle cell heterozygous, Sickle cell homozygous, HbE heterozygous and HbD Iran heterozygous and compound heterozygous for HbQ and β thalassemia. One case each of HbJ Meerut, Compound heterozygous for HbS, HbE and HbJ with β thalassemia was diagnosed along with Compound heterozygous for HbS and Delta β thalassemia which is in concordance with studies conducted by Mondal et al,[93] Shrivastav et al,[92] Singh et al[81] and Pachori et al[85] in which Beta thalassemia trait was diagnosed to be the commonest Hb abnormality. Shrivastav et al[92] study found Beta thalassemia trait to the commonest Hb abnormality followed by thalassemia major, HbS heterozygous, Hb S homozygous, Compound heterozygous for HbS and beta thalassemia, HbD Punjab heterozygous, HbD Punjab homozygous and HbE heterozygous, HbE homozygous, Compound heterozygous for HbD and beta thalassemia in a decreasing frequency It is in contrast to study conducted by Iqbal et al[78] where Sickle cell trait followed by Beta

thalassemia trait and sickle cell homozygous. In a study conducted by Goswami et al[89] HbE trait was diagnosed as the commonest Hb abnormality followed by HbE homozygous and Beta thalassemia trait in a decreasing frequency.

1) BETA THALASSEMIA

a) Beta thalassemia trait (BTT)

In the present study, Of the 255 cases majority were in the age group of 21 to 30 years which is in concordance with study conducted by Pachori et al[85] where majority of cases were in the age group of 16 to 45 years. Female preponderance (M:F= 1:1.6) was seen which is similar to studies conducted by Goswami et al.[89] however in contrast Pachori et al[85] had more males in their study.

Diagnosis

In present study HbA2 ranged from 4% to 10.2% which is similar to studies conducted by Khera et al[77] where HbA2 ranged from 4% to 8.2%.

In the present study mean value of HbA2 was 5.7% which is in concordance with the study conducted by Khera et al,[77] Shrivastav et al[92] and Iqbal et al[78] where mean value for HbA2 was 5.7%, 5.3% and 5.2% respectively. HbF ranged from 0.2% to 16% which is in contrast to studies conducted by Khera et al [77] where HbF ranged between 0.2 to 4.9%. Mean value of HbF was 1.6% which is in concordance with the study conducted by Khera et al[77] and Shrivastav et al [92] where mean value of HbF was 1.5% and 1.7% respectively however slightly higher mean value of HbF (2.3%) was reported by Pachori et al[85]. (Table 17)

HbA ranged 63.6% to 95.9% which is similar to studies conducted by Khera et al[77] where HbA ranged between 71.7% to 89.1%. Mean value of HbA was 86.15% which is in concordance with the study conducted by Shrivastav et al[92] and Khera et al[77] where mean value of HbA was 83.3% and 83.6% respectively.

Table 17: Comparison of Abnormal HPLC findings with other studies in Beta-Thalassemia trait

Abnormal HPLC finding	Pachori et al [85] (North India)	Chandra Shekhar et al [82](South India)	Shrivastav et al (92) (Western India)	Present study
Mean HbA2 (%)	5.1	5.4	5.3	5.7
Mean HbF (%)	2.3	1.4	1.7	1.5

Clinical Profile

In present study pallor was noted in 54.1% cases, Icterus in 3.1% cases and splenomegaly in 1.1% cases.

Diagnosis

In our study Hb levels ranged from 3.3 to 14.9 g/dl which is in concordance with the study conducted by Khera et al[77] where Hb ranged from 3.2 to 14.4 g/dl and 7.6 to 14.7 g/dl. Mean value of Hb was 10.3 g/dl which is similar to study conducted by Shrivastav et al,[92] Mujawar et al,[97] Iqbal et al [78] where mean value of Hb was 10.4, 9.9 and 9.8 g/dl respectively. It is contrast to studies conducted by Bhalodia et al[80] and Khera et al[77] where mean value of Hb was 8.3 and 9.3 g/dl respectively.

In present study majority cases had anemia (80%) which is similar to studies conducted by Singh et al[81] and Bhalodia et al[80] which shows anemia in 83.2% and 72% respectively and majority cases show moderate anemia (56.2%) which is in contrast to studies conducted by Singh et al[81] where majority cases show severe anemia. On peripheral examination majority cases showed anisocytosis microcytosis, hypochromia and target cells which is similar to study conducted by Bhalodia et al.[80]

In our study RBC count ranged from 1.51 to 7.55x10^6/µL and mean value was 5.17x10^6/µL which is comparable with the study conducted by Khera et al[77] where RBC count ranged from 1.65 to 7.12x10^6/µL and mean value was 5.18x10^6/µL. It is contrast to studies conducted by Bhalodia et al[80] and Mujawar et al[97] where mean value was 4.39x10^6/µL in both studies. In present study 43% cases had normal range RBC count i.e. 4.5 -5.5x10^6/µL which is in contrast to studies conducted by Bhalodia et al[80] and Singh et al[81] where 42.3% and 57.1% cases respectively had high RBC count.

In present study MCV ranged from 46 to 119 fl and mean value was 65.8 fl which is similar to study conducted by Khera et al[77] where MCV ranged from 49.8 to 85.2 fl and mean value was 62.7 fl. In our study 95.2% cases had low MCV (<80fl) which is comparable to studies conducted by Singh et al[81] and Bhalodia et al[80] where All cases had low MCV (<80fl). In the present study Mentzers' s Index was found to be less than 13 in 62.4% cases which is comparable to 82.8% cases in study conducted by Sarvaiya et al.[95]

In our study MCH ranged from 11.8 to 39.9 pg which is in contrast to study conducted by Khera et al[77] where MCH ranged from 10.9 to 27.2 pg. Mean value of MCH was 20.6 pg which is in concordance with studies conducted by Iqbal et al,[78] Bhalodia et al[80] and Khera et al[77] where mean value of MCH was 20.8 pg, 19.4pg and 19.6pg respectively. In our study 94.5% cases had low MCH (<27 pg) which is comparable to studies conducted by Singh et al[81] and Bhalodia et al[80] where All cases had low MCH.

In the present study MCHC ranged from 19.9 to 34.3 g/dl and mean value was 30.6 g/dl which is in concordance with the study conducted by Khera et al[77] where MCHC ranged from 20.9 to 36 g/dl and mean value was 31.4 g/dl. In our study majority cases (91%) had low MCHC (<32g/dl) which is in contrast to studies conducted by Bhalodia et al[80] and Singh et al[81] where 69.2% and 83.3% cases respectively had normal range MCHC (32-37 g/dl).

In present study RDW ranged from 13.6% to 35% and mean value was 17.6% which is in concordance with the study conducted by Khera et al [77] where RDW ranged from 14.6 to 36.7% and mean value was18.7%. (Table 18)

Table 18: Comparison of different hematological parameters with other studies in Beta-thalassemia trait.

Hematological Parameters (Mean value)	Pachori et al [85] (North India)	Chandra Shekhar et al [82] (South India)	Shrivastav et al [92] (Western India)	Present study
Hb (g/dl)	9.1	10.4	10.4	10.3
RBC(10^6/µL)	5.7	5.2	5.4	5.1
MCV (fl)	69.1	65.5	62.1	65.8
MCH (pg)	21.3	19.9	19.4	20.6
MCHC (g/dl)	27.0	30.5	30.3	30.6
RDW (%)	15.8	16.5	18.7	17.6

In the present study Molecular genetics in beta thalassemia trait cases found IVS1-5 (G->C) to be the most common mutation to be encountered which is in concordance with the studies conducted by Chatterjee et al[98] and Nadkarni et al.[90]

b) Beta thalassemia Homozygous

In the present study, Of the 22 cases no sex preponderance was noted which is in concordance with the studies conducted by Bhokare et al[99] and Goswami et al.[89] It is in contrast to studies conducted by Pachori et al[85] where male preponderance was noted and Shrivastav et al[92] where slight female preponderance was noted.

Diagnosis

In our study HbF ranged from 20.1 to 97% which is in concordance to study conducted by Khera et al[77] where HbF ranged between 30.6 to 91% and the mean value of HbF was 68.4% which is in concordance with the studies conducted by Tripathi et al[100] and Pachori et al[85] where

mean value of HbF was 64.1% and 72.4% respectively. In present study HbA2 ranged from 0.6% to 8.2% and mean value was 2.9% which is similar to studies conducted by Khera et al[77] where HbA2 ranged from 0% to 4.5% and mean value was 2.8%. (Table 19)

In our study HbA ranged 0.4% to 72% which is similar to studies conducted by Khera et al[77] where HbA ranged between 4.6% to 60.5% and mean value of HbA was 27.2% which is in concordance with the study conducted by Shrivastav et al,[92] Tripathi et al[100] and Khera et al[77] where mean value of HbA was 21.2%, 24.6% and 25.0% respectively.

Table 19: Comparison of Abnormal HPLC findings with other studies in Beta-Thalassemia homozygous

Abnormal HPLC Finding	Pachori et al [85](North India)	Chandra Shekhar et al [82] (South India)	Shrivastav et al [92] (Western India)	Present study
Mean HbF (%)	72.4	88	78.6	68.4
Mean HbA2 (%)	2.7	3.1	3.8	2.9

Clinical features

In present study pallor was noted in 72.1% cases, Icterus in 63.6% cases and splenomegaly in 91% cases which is in comparable to study conducted by Bhokare et al[99] where pallor was noted in 88.8% cases, Icterus in 72.2% cases and splenomegaly in 83.3% cases.

Hematological parameters

In our study Hb levels ranged from 2.7 to 10.1 g/dl which is comparable to the study conducted by Shankar et al[94] where Hb ranged from 1.3 to 9 g/dl. Mean value of Hb was 6.4 g/dl which is similar to study conducted by Mujawar et al[97] where mean value of Hb was 5.9 g/dl.

In our study on peripheral examination majority cases showed marked anisopoikilocytosis, microcytosis, hypochromia and nucleated RBCs which is similar to studies conducted by Bhalodia et al.[80]

In our study RBC count ranged from 1.01 to 4.6x10^6/µL which is comparable with the study conducted by Khera et al[77] where RBC count ranged from 0.79 to 4.3x10^6/µL. The mean value of RBC count was 2.6x10^6/µL which is in concordance with the study conducted by Shrivastav et al[92] and Mujawar et al[97] where mean value was 2.4x10^6/µL and 2.8x10^6/µL respectively. In present study MCV ranged from 62 to 94 fl which is in concordance with the study conducted by Khera et al[77] where MCV ranged from 56.7 to 79.7 fl. Mean value of MCV was 73.0 fl which is concordance with the study conducted by Tripathi et al[100] and Mujawar et al [97] where mean for MCV was 68.7 fl and 71.2 fl.

In our study MCH ranged from to 19.9 to 32.9 pg which is comparable to study conducted by Khera et al[77] where MCH ranged from 17.7 to 22.3 pg. Mean value of MCH was 24.9 pg which is in concordance with study conducted by Shrivastav et al[92] where mean was 23.2pg. MCHC ranged from 26 to 44 g/dl which is in concordance with study conducted by Khera et al[77] where MCHC ranged from 23.8 to 34 g/dl. Mean value of MCHC was 32.1 g/dl in our study which is similar to studies conducted by Khera et al[77] and Deb et al [101] where mean value of MCHC was 31.0 and 34.1 g/dl respectively.

In present study RDW ranged from 19% to 37% which is in concordance with the study conducted by Khera et al[77] where RDW ranged from 20.2 to 36.2%. Mean value of RDW was 28.0% which is in concordance with the study conducted by Iqbal et al[78] where mean value of RDW was 29.3%. (Table 20)

Table 20: Comparison of different hematological parameters with other studies in Beta-thalassemia homozygous.

Hematological Parameters (Mean value)	Pachori et al[85](North India)	Chandra Shekhar et al [82](South India)	Shrivastav et al[92] (Western India)	Present study
Hb (g/dl)	8.0	4.8	5.3	6.4
RBC(10^6/µL)	4.1	2.4	2.4	2.6
MCV (fl)	62.6	67.9	68.5	73.0
MCH (pg)	19.2	21.1	23.2	25.0
MCHC (g/dl)	24.3	31	34.1	32.2
RDW (%)	22.3	30.9	23.4	28.1

2) HbD Punjab heterozygous

In the present study, Of the 38 cases age group ranged from one year to 54 years which is comparable to study conducted by Srinivas et al[102] where age group ranged from 6 to 61 years. Female preponderance was noted which is similar to study conducted by Shrivastav et al.[92] It is in contrast to studies conducted by Srinivas et al[102] where male preponderance was noted.

Diagnosis

In our study HbD ranged from 11 to 40.4% and the mean value was 33.8% which is in concordance to study conducted by Khera et al[77] where HbD ranged between 13.4 to 38.5% and mean value was 34.8%. HbF ranged from 0 to 1.6% and mean value was 0.7% which is in concordance to study conducted by Khera et al[77] where HbF ranged between 0.2 to 1.2% and the mean value was 0.6%. HbA2 ranged from 1.7% to 3.5% which is comparable to study conducted by Khera et al[77] where HbA2 ranged from 0.7% to 1.8% and mean value of HbA2 was 2.7% which is in concordance with the study conducted by Tripathi et al[100] and Shrivastav et al[92] where mean value for HbA2 was 2.3 and 2.4% respectively. HbA ranged from 49.8% to 85.3% which is similar to studies conducted by Khera et al[77] where HbA ranged between 50.7%

to 77% and mean value of HbA was 60.5% which is comparable with the study conducted by Tripathi et al[100] where mean value of HbA was 56.6%. (Table 21)

Table 21: Comparison of Abnormal HPLC findings with other studies in HbD Punjab heterozygous.

Abnormal HPLC Finding	Pachori et al [85](North India)	Chandra Shekhar et al[82] (South India)	Shrivastav et al[92] (Western India)	Present study
Mean HbD (%)	31.0	35.4	37	33.8
Mean HbF (%)	2.0	2.1	0.6	0.6
Mean HbA2 (%)	2.7	2.5	2.4	2.7

Clinical profile

In present study pallor was noted in 36.8% cases, Icterus in 2.6% cases which is in contrast to study conducted by Srinivas et al[102] where such cases were clinically asymptomatic.

Hematological parameters

In our study Hb levels ranged from 3.9 to 16.3 g/dl which is in comparable to the study conducted by Khera et al[77] where Hb ranged from 5.2 to 15.8 g/dl. Mean value of Hb was 11.0 g/dl which is in concordance to study conducted by Shrivastav et al[92] and Srinivas et al[102] where mean value of Hb was 11 and 11.8 g/dl respectively. On peripheral examination majority cases showed microcytosis, hypochromia and few target cells which is similar to study conducted by Bhalodia et al.[80]

In our study RBC count ranged from 1.38 to 6.07x10^6/µL and mean was 4.1x10^6/µL which is comparable with the study conducted by Khera et al.[77] In present study MCV ranged from 52.1 to 112 fl and mean was

83.3 fl which is in concordance with the study conducted by Khera et al[77] where MCV ranged from 47.2 to 102 fl and mean was 78.4 fl.

In our study MCH ranged from 13 to 36.7 pg which is comparable to study conducted by Khera et al[77] where MCH ranged from 12.1 to 30.9 pg. Mean value of MCH was 27.11 pg which is comparable with study conducted by Srinivas et al[102] where mean value was 25.0 pg. MCHC ranged from 24.6 to 36.7 g/dl which is in concordance with the study conducted by Khera et al[77] where MCHC ranged from 25.6 to 35.8 g/dl. Mean value of MCHC was 32.3 g/dl which is similar to studies conducted by Srinivas et al[102] Khera et al [77] and Shrivastav et al [92] where mean value of MCHC was 31.0, 31.5 and 32.3 g/dl respectively.

In present study RDW ranged from 12.9% to 35.7% which is in concordance with the study conducted by Khera et al[77] where RDW ranged from 13.9 to 26.8%. (Table 22)

Table 22: Comparison of different hematological parameters with other studies in HbD Punjab heterozygous.

Hematological Parameters (Mean value)	Pachori et al[85](North India)	Chandra Shekhar et al [82] (South India)	Shrivastav et al[92] (Western India)	Present study
Hb (g/dl)	8.7	10	11	11
RBC(10^6/µL)	3.5	3.6	4.7	4.1
MCV (fl)	68.9	82.6	76.8	83.3
MCH (pg)	20.5	27.3	24.7	27.1
MCHC (g/dl)	24.5	33	32.3	32.3
RDW (%)	16.6	17.2	19.2	17.8

3) **HbQ India heterozygous**

In the present study, Six cases age group ranged from 24 to 53 years which is comparable to study conducted by Dhawle et al[103] where age group ranged from 3 to 60 years. In present study female preponderance was noted which is in contrast to study conducted by Dhawle et al[103] where male preponderance was noted.

Diagnosis

HbQ ranged from 15.3 to 20.3% which is in concordance to study conducted by Dhawle et al[103] HbQ ranged between 15.6 to 17.4%. HbF ranged from 0 to 0.8% which is in concordance to study conducted by Dhawle et al[103] where HbF ranged between 0.2 to 1.4%. HbA2 ranged from 2.1% to 3.7% which is comparable to study conducted by Dhawle et al[103] HbA2 ranged from 1.2% to 6.0%. HbA ranged 67.9% to 81.9% which is similar to studies conducted by Dhawle et al [103] HbA ranged between 71.1% to 76.1%.

Hematological parameters

Hb levels ranged from 8.3 to 12.9 g/dl and RBC count ranged from 3.98 to 4.81x10^6/µL which is comparable to the study conducted by Dhawle et al[103] where Hb ranged from 10.3 to 15.2 g/dl and RBC count ranged from 4.39 to 5.39x10^6/µL. MCV ranged from 68.6 to 93.5 fl which is in concordance with the study conducted by Dhawle et al[103] where MCV ranged from 62.2 to 100 fl.

In our study MCH ranged from 20.8 to 30.6 pg and MCHC ranged from 30.3 to 33.7 g/dl which is comparable to study conducted by Dhawle et al[103] where MCH ranged from 19.1 to 31.8 pg and MCHC ranged from 29.8 to 33.2 g/dl. RDW ranged from 12.0% to 18.7% which is in concordance with the study conducted by Dhawle et al[103] where RDW ranged from 13.2 to 15.6%.

4) Compound Heterozygous for HbD Punjab & β Thalassemia

In the present study, Of the four cases age group ranged from 28 to 72 years which is in contrast to study conducted by Srinivas et al[102] where age group ranged from 9 to 19 years. No sex preponderance was noted which is in contrast to study conducted by Srinivas et al[102] where male preponderance was noted.

Diagnosis

In our study mean value of HbD and HbA2 was 70.1% and 4.7% respectively which is in contrast to study conducted by Srinivas et al[102] where mean value of HbD and HbA2 was 53.8% and 8.2% respectively. Mean value of HbF and HbA was 2.2% and 21.5% respectively which is in contrast to study conducted by Srinivas et al[102] where mean value of HbF and HbA was 5.8% and 8.2% respectively.

Hematological parameters

Mean value of Hb and RBC count was 11.82 g/dl and $5.36x10^6$/µL respectively in present study. It is in contrast to study conducted by Srinivas et al[102] where mean value of Hb and RBC count was 6.9 g/dl and $3.46x10^6$/µL respectively. Mean value of MCV was 69.8 fl. It is in concordance to study conducted by Srinivas et al[102] where mean value of MCV was 69.6 fl.

Mean value of MCH and MCHC was 22.27 pg and 31.7 g/dl respectively. It is in contrast to study conducted by Srinivas et al[102] where mean value of MCH and MCHC was 16.9 pg and 21.9 g/dl respectively.

5) Sickle Cell Heterozygous

In present study of two cases diagnosed female preponderance was noted. It is in contrast to studies conducted by Shrivastav et al[92] and Goswami et al[89] where no sex preponderance was noted.

Diagnosis

In present study, HbS was 29.9% and 31.1% respectively which is similar to study conducted by Shankar et al.[94] HbF was 0.8% and 1.6% which is similar to study conducted by Shrivastav et al.[92] HbA2 was 3.1% and 3.2% which is comparable to study conducted by Shrivastav et al.[92] HbA was 58.6% and 64.2% which is comparable to studies conducted by Shankar et al[94] and Shrivastav et al.[92]

Hematological parameters

In present study, Hb was 9.1 and 10.9 g/dl respectively which is comparable to study conducted by Shrivastav et al.[92] RBC count was 3.82 and 4.52x10^6/µL which is similar to studies conducted by Shrivastav et al.[92] In our study MCV, MCH and MCHC was 68 and 87 fl, 20.2 and 28.2 pg and 29.8 ad 32.7 g/dl respectively which is comparable to study conducted by Shrivastav et al.[92] RDW was 14.1% and 17.7% respectively which is in contrast to study conducted by Shrivastav et al.[92]

6) Sickle Cell Homozygous

In present study no sex preponderance was noted. It is in contrast to studies conducted by Shrivastav et al[92] and Goswami et al[89] where female preponderance was noted.

Diagnosis

In present study, HbS was 66% and 71% respectively which is similar to study conducted by Tripathi et al[100] and Shrivastav et al.[92] HbF was 17.1% and 24.5% which is similar to study conducted by Shrivastav et al[92] and Tripathi et al.[100] HbA2 was 1.6% and 3.5% which is comparable to study conducted by Tripathi et al[100] and Shrivastav et al.[92] HbA was 5.3% and 8.4% which is comparable to study conducted by Shrivastav et al.[92]

Hematological parameters

In present study on peripheral smear there was moderate to severe degree of anisopoikilocytosis with mild hypochromia, sickle cells, few target cells which is in concordance with the study conducted by Shankar et al.[94] Hb was 8.7 and 11.9 g/dl respectively which is comparable to study conducted by Tripathi et al.[100] RBC count was 2.9 and 4.3x10^6/µL which is similar to studies conducted by Shrivastav et al.[92] MCV, MCH and MCHC was 84 and 93 fl, 27.5 and 30.1 pg and 32.3 and 32.7 g/dl

respectively which is comparable to study conducted by Shrivastav et al.[92]

7) HbE heterozygous

In present study no sex preponderance was noted. It is in concordance to study conducted by Shrivastav et al[92] where no sex preponderance was noted.

Diagnosis

In present study, HbA2 was 24.5% and 32.8% respectively which is similar to study conducted by Khera et al.[77] HbF was 0% and 0.7% which is similar to study conducted by Shrivastav et al.[92]

Hematological parameters

In present study, Hb was 10.9 and 13.7 g/dl respectively which is comparable to study conducted by Shrivastav et al.[92] RBC count was 4.32 and 5.32x10^6/µL which is similar to studies conducted by Shrivastav et al.[92] RDW in our study was 16.6 and 25.3% which is comparable to study conducted by Khera et al.[77]

8) HbD Iran heterozygous

Diagnosis

In our study, HbA2 was 40.9% and 44.5% respectively which is similar to study conducted by Bhalodia et al.[80]

9) Compound heterozygous for HbQ India and Beta thalassemia

In present study, diagnosed no sex preponderance was noted. It is in concordance to study conducted by Phanasgaonkar et al[104] where no sex preponderance was noted.

Diagnosis

In present study, HbQ was 7.8% and 8.2% respectively which is similar to study conducted by Harrison et al.[87] HbA2 was 5.5 and 6.7% which

is comparable to study conducted by Harrison et al.[87] HbA was 81.7% and 82.6% which is comparable to study conducted by Harrison et al.[87]

Hematological parameters

In present study, RBC count was 4.88 and 5.49x10^6/µL respectively which is similar to studies conducted by Harrison et al.[87] In our study MCV, MCH and MCHC was 64.5 and 71.1 fl, 20.6 and 21.9 pg and 31.9 and 38.8 g/dl respectively which is comparable to study conducted by Harrison et al.[87]

10) Compound heterozygous for HbS and Beta Thalassemia

One case was diagnosed which was male

Diagnosis

In present study, HbS was 34.9% which is in contrast to study conducted by Shankar et al.[94] HbF was 35.4% which is in contrast to study conducted by Khera et al.[77] HbA2 was 4% which is in contrast to study conducted by Shankar et al.[94] HbA was 18.3% which is in contrast to study conducted by Shankar et al.[94]

Hematological parameters

In present study, Hb was 2.4 g/dl which is in contrast to study conducted by Shankar et al.[94] RBC count was 0.93x10^6/µL which is in contrast to study conducted by Shrivastav et al.[92] In our study MCV, MCH and MCHC was 87.1fl, 26 pg and 29.9 g/dl respectively which is comparable to study conducted by Shrivastav et al.[92] RDW was 24.7% which is comparable to study conducted by Shrivastav et al.[92]

11) Compound heterozygous for HbE and Beta Thalassemia

One case was diagnosed which was female

Diagnosis

In present study, HbA2 was 61.1% which is similar to study conducted by Khera et al.[77] HbF was 31.7% which is comparable to study conducted by Tripathi et al.[100]

Hematological parameters

In present study, Hb was 4.5 g/dl which is in concordance with the study conducted by Khera et al. [77] RBC count was 2.54x10^6/µL which is in concordance with study conducted by Khera et al.[77] In our study MCV, MCH and MCHC was 62 fl, 17.7 pg and 28.5 g/dl respectively which is comparable to study conducted by Tripathi et al. [100] RDW in our study was 34.8% which is comparable to study conducted by Khera et al.[77]

12) HbJ Meerut

One case was diagnosed which was female.

Diagnosis

In present study, P3 was 7.4% which is in contrast with study conducted by Pant et al.[105] HbA2 was 3.3% and HbA was 89.3% which is comparable to study conducted by Pant et al.[105]

Hematological parameters

In present study, Hb was 8.8 g/dl which is in contrast to the study conducted by Pant et al.[105] RBC count was 3.41x10^6/µL which is similar with study conducted by Pant et al.[105] In our study MCV, MCH and MCHC was 77.3 fl, 25.7 pg and 33.2 g/dl respectively which is in contrast to study conducted by Pant et al.[105]

13) Compound heterozygous for HbJ and Beta thalassemia

One case was diagnosed and was female. This was a rare entity where in our study on HPLC P3 peak was 23.4% at a retention time of 1.57 minute. Various abnormal hemoglobin that causes spurious increase in

P3 were Hb Cambden, Hb Hope, HbJ Oxford and Hb Austin. HbA2 was 4.4%, HbF was 2.1% and HbA was 70.1%. In present study Hb levels were 10.0 g/dl. RBC count was $5.12x10^6$/µL. MCV, MCH and MCHC was 91 fL, 28.3 pg and 31 g/dl respectively. RDW was 31.2%.

14) Compound heterozygous for HbS and delta Beta thalassemia

One case was diagnosed which was male

Diagnosis

In present study, HbS was 50.6%, HbF was 29.1%, HbA2 was 3.2% and HbA was 2.2%.

15) Borderline cases

HbA2 interpretation plays an important role in the screening programs for Beta thalassemia in which if HbA2>4%, the patient would be labelled as Beta thalassemia trait. Individuals with borderline HbA2 were difficult to be classified based on hematological parameters alone.[106] These cases were commonly encountered in daily HPLC reporting where HbA2 was between 3.6% to 3.9% and constituted around 16% in present study which is similar to population in the countries where thalassemia is common and ranged from 2.2% to 16.7%.[107] Particular care has to be paid for the interpretation of borderline HbA2 in which the HbA2 level above the normal reference interval but not in the classical range of beta thalassemia and required further investigation before reaching out on conclusion. Studies have suggested that Iron deficiency anemia can mildly reduce HbA2 and Megaloblastic anemia can mildly increase HbA2.[108-111] So, in these cases we recommend serum iron, Vit B12 and folate levels and repeat HPLC after adequate treatment. In present study we had 65 cases which had borderline HbA2 levels out of which 3 cases had high HbF and molecular genetics were recommended to them, of which 2 cases had genetics. One was positive for IVS1-1 and other for Fr8/9 mutations respectively. These 2 cases were then diagnosed under

Beta thalassemia trait. Rest of the cases were out of follow up. This was similar to study conducted by Paglietti et al[112] and Rosnah B et al. [106]

SUMMARY AND CONCLUSION

The present study was conducted in the Department of Pathology at Dayanand Medical College and Hospital, Ludhiana, from 1st January, 2018 to 30th June, 2020. Out of 3135 samples received, 405 samples were analyzed and relevant data was compiled. The following features were highlighted in the study

- The age of the patients ranged from one month to 72 years, with a mean age of 28.5 years. Majority were in age group of 21 to 30 years (181 cases, 44.5%) and were females (268 cases, 66.2%), with a male to female ratio of 1:1.9.
- Majority of the patients were diagnosed as having β-thalassemia trait (63%), followed by HbD Punjab heterozygous (9.3%), β-thalassemia homozygous (5.4%), HbQ India heterozygous (1.5%), Compound heterozygous for HbD Punjab and Beta thalassemia (1%). 2 cases each were diagnosed as Sickle cell heterozygous, Sickle cell homozygous, HbE heterozygous and HbD Iran heterozygous, Compound heterozygous for HbQ and β thalassemia. One case each of HbJ Meerut, Compound heterozygous for HbS, HbE and HbJ with β thalassemia was diagnosed along with Compound heterozygous for HbS with delta β thalassemia. Also there were 65 cases (16.1%) which was diagnosed as borderline
- Beta-thalassemia trait (63% of total cases) had HbA2 between 4 to 6.9% (96%) with mean of 5.6%, and HbF less than 5% with a mean of 1.6%. Majority cases (56.2%) had moderate anemia degree(mean Hb 10.3 g/dl). Majority cases had low MCV< 80 fl (95.2%), Mentzer's index<13 (62.4%) with a mean of 13.4, low MCH < 27 pg (94.5%), low MCHC< 32 g/d (91.0%) and RDW between 15 to 20%(82.3%). IVS1-5 (G->C) mutation (28.6%)was the commonest.
- Beta thalassemia homozygous (5.4% of total cases) with M:F ratio of 1:1. Majority had increased HbF (HbF>80%) (50.0%), HbA2 less than 4% (86.4%) with mean of 2.9%. Majority had

anemia of severe degree (56.%) with a mean Hb of 6.4g/dl, low MCV< 80 fl (86.3%) with a mean of 73.0fL, low MCH < 27 pg (63.6%) , low MCHC< 32 g/d (59.1%) . Fr-8/9 (+G) mutation (42.9%) was the commonest.

- After beta thalassemia HbD Punjab heterozygous (9.3%) was commonest. Majority had HbD≥30% (86.8%), with a mean of 33.8% and retention time ranged between 3.83 to 3.89 minute. Majority cases had normal MCV (63.1%), normal MCH (55.3%), normal MCHC (71.1%)
- Borderline cases constituted 16.1%. All cases had HbA2 between 3.6% to 3.9% on HPLC. Majority cases had normal Hb and low RBC count and normal MCV, MCH and MCHC.

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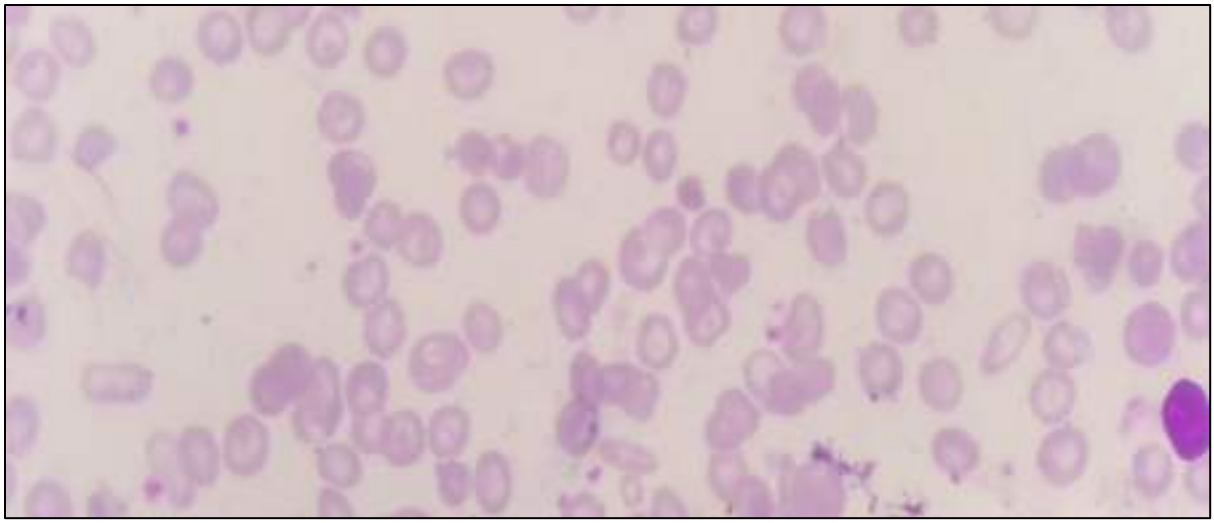
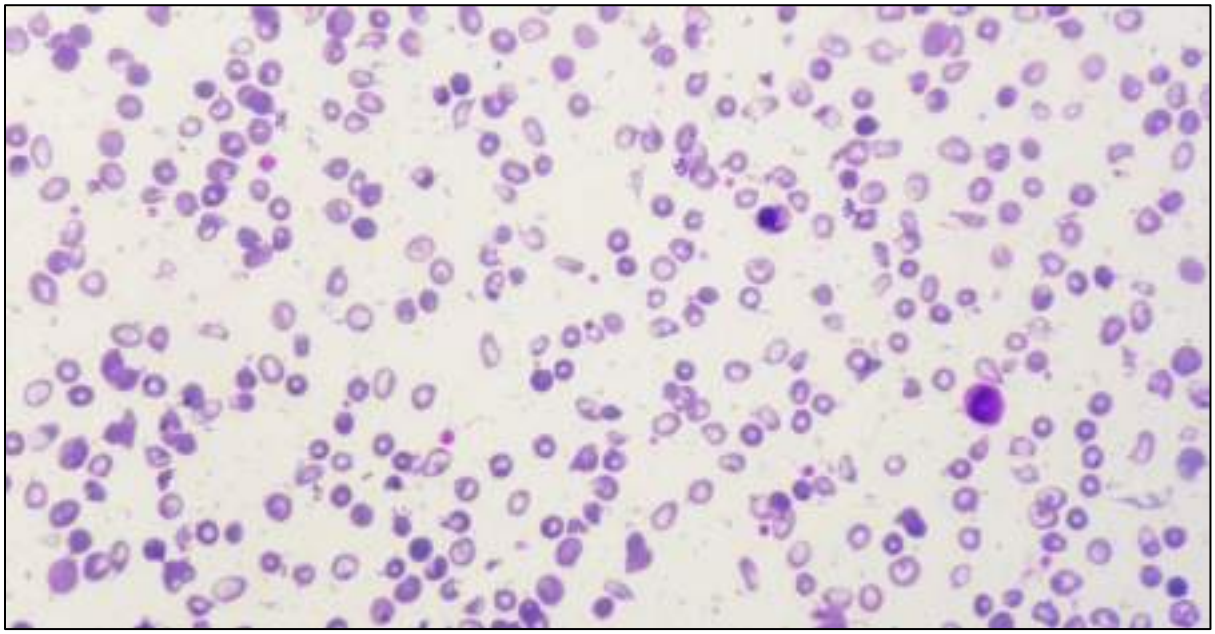
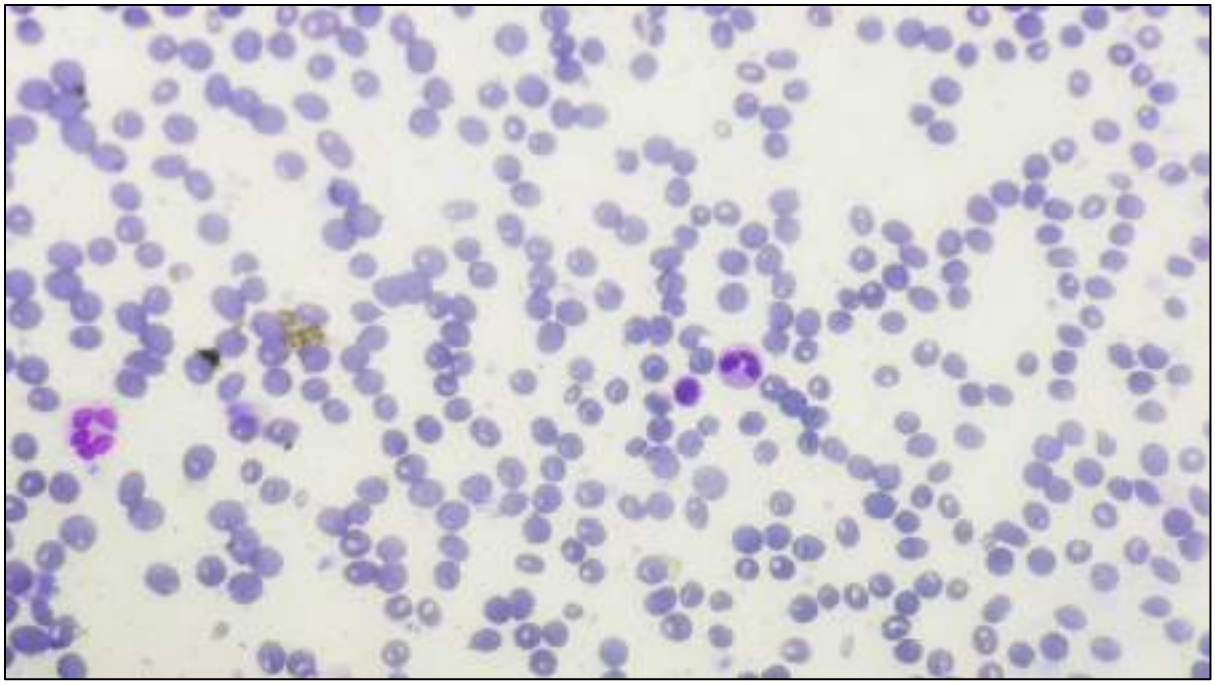


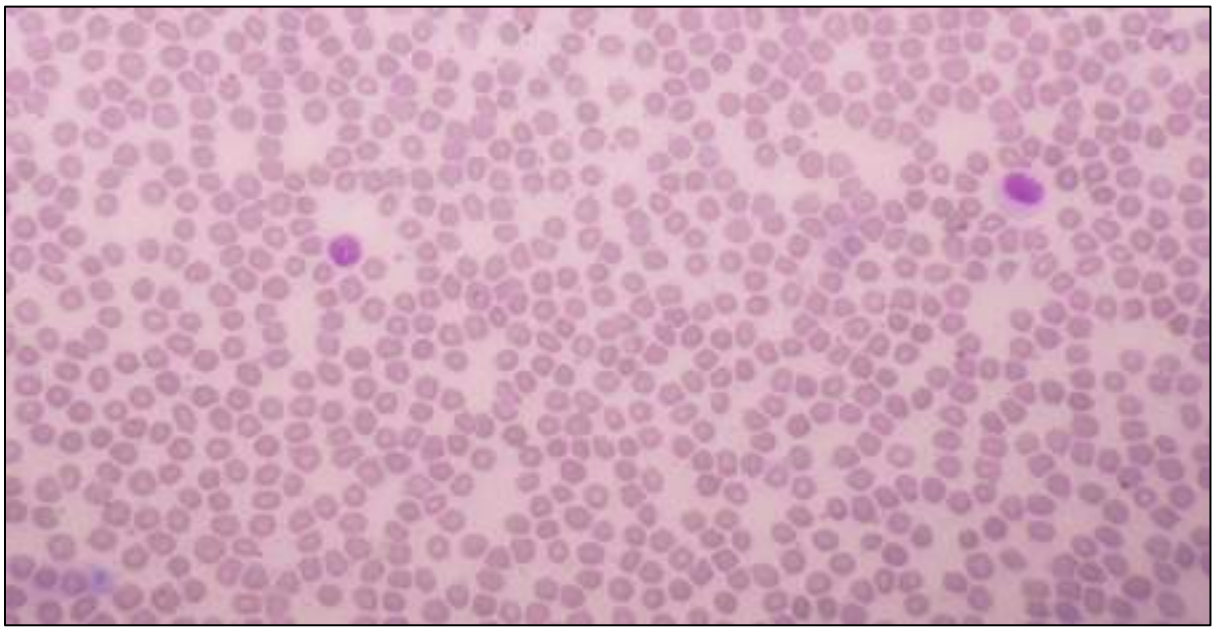
Picture 1: Peripheral Blood smear showing mild degree of anisocytosis with microcytic hypochromic RBC picture and few target cells in β-thalassemia heterozygous (Giemsa400X)



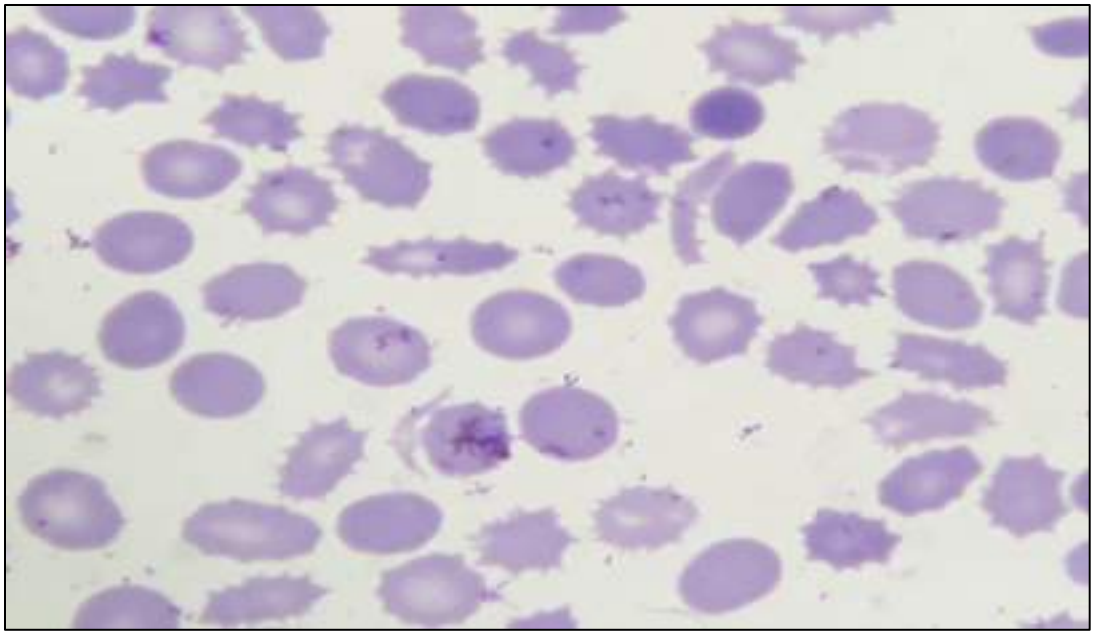
Picture 2: Peripheral Blood smear showing marked degree of anisopoikilocytosis with microcytic hypochromic RBC picture, few tear drop cells and target cells along with nucleated red cells in β-thalassemia homozygous (Giemsa 400X)



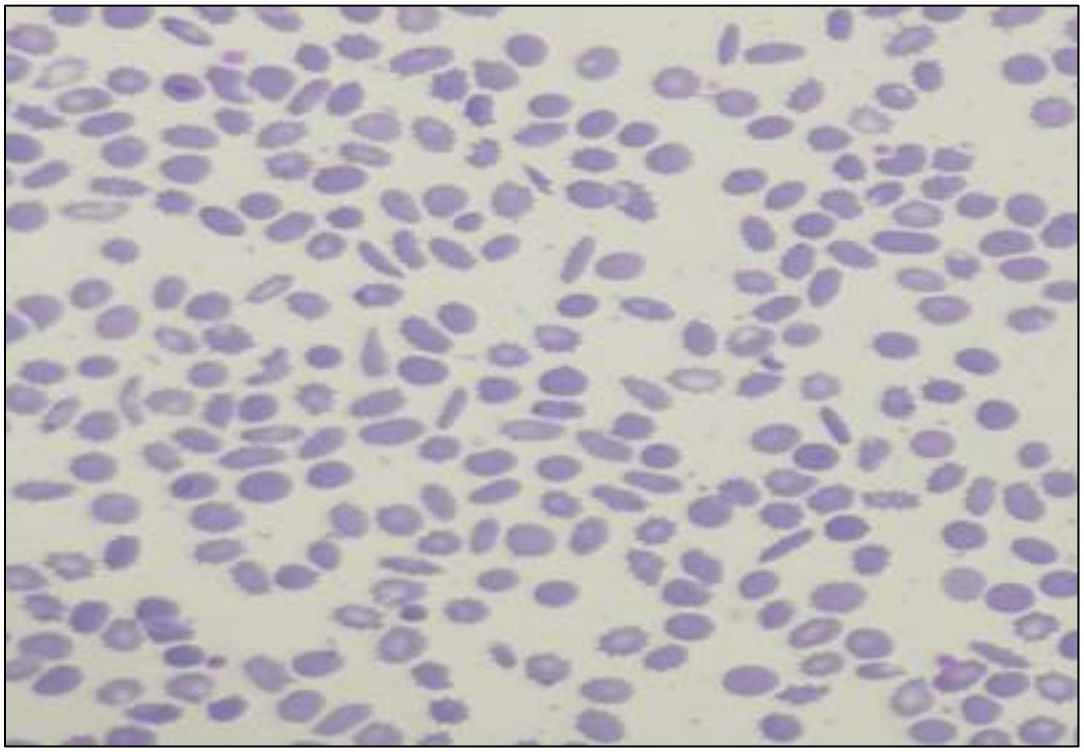
Picture 3: Peripheral Blood smear showing microcytosis, hypochromia with few target cells in HbD Punjab heterozygous (Giemsa 400X)



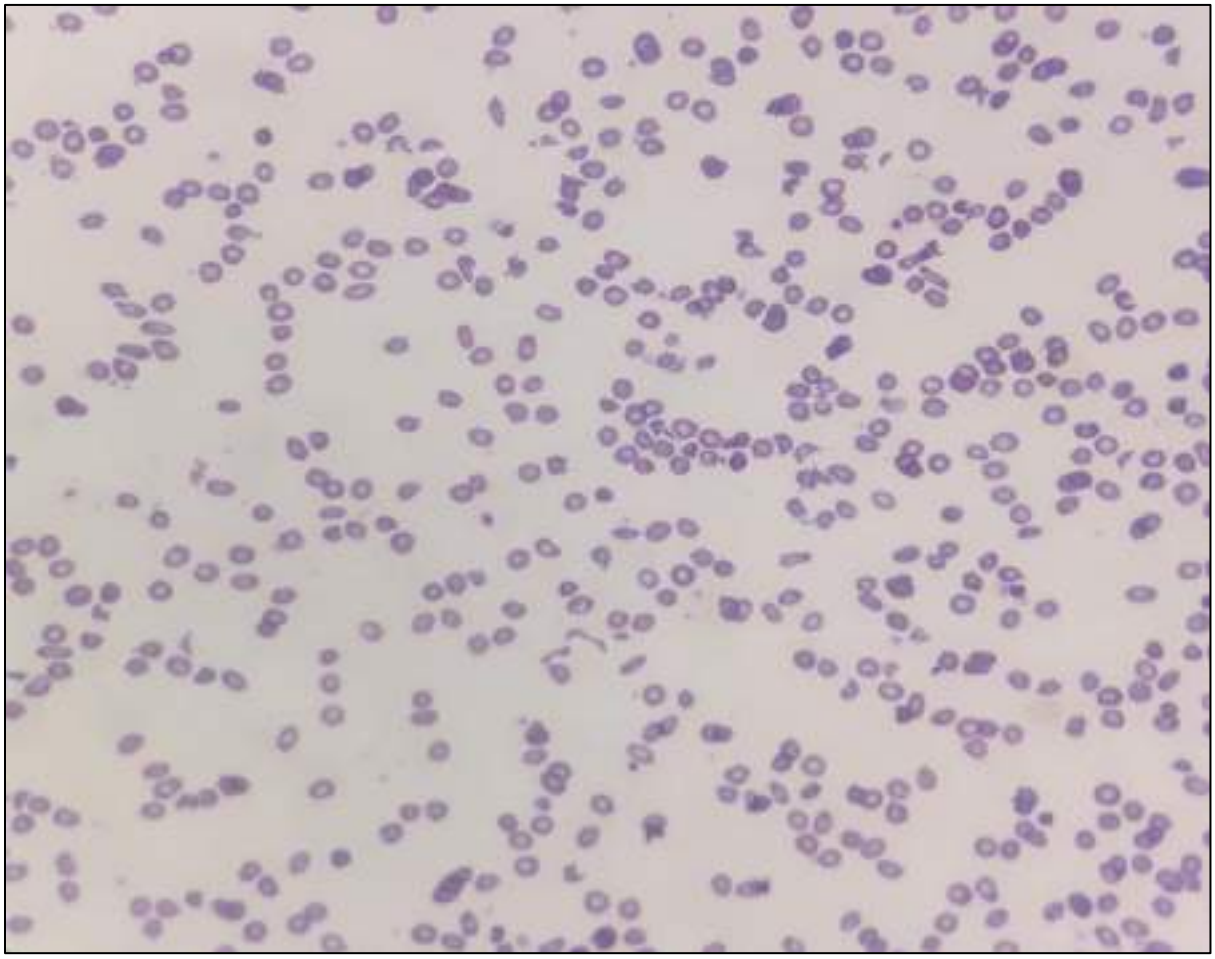
Picture 4: Peripheral Blood Smear showing Normocytic normochromic red blood cell picture in HbQ India heterozygous.(Giemsa 400X)



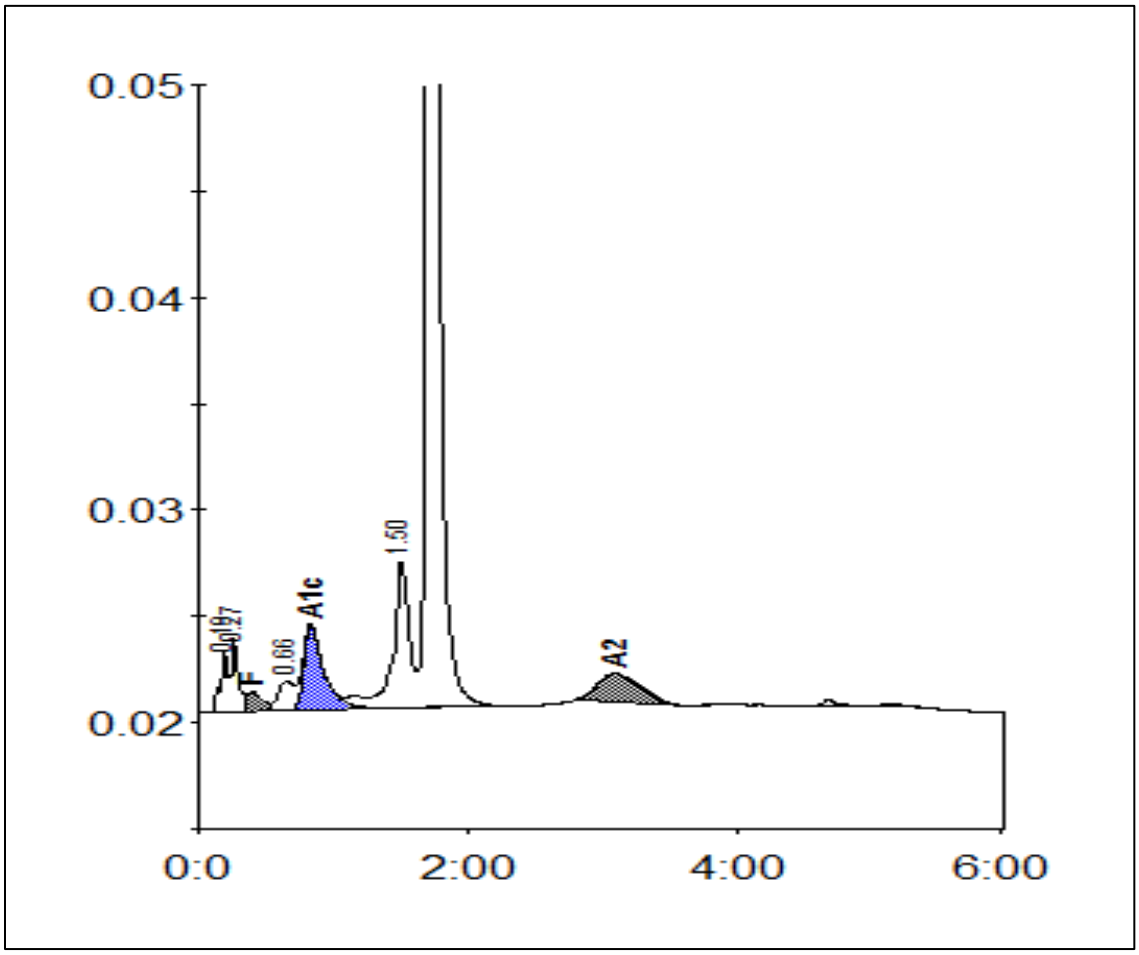
Picture 5: Peripheral Blood smear showing Normocytic normochromic RBC picture with few sickle cells in Sickle cell heterozygous (Giemsa 1000X)



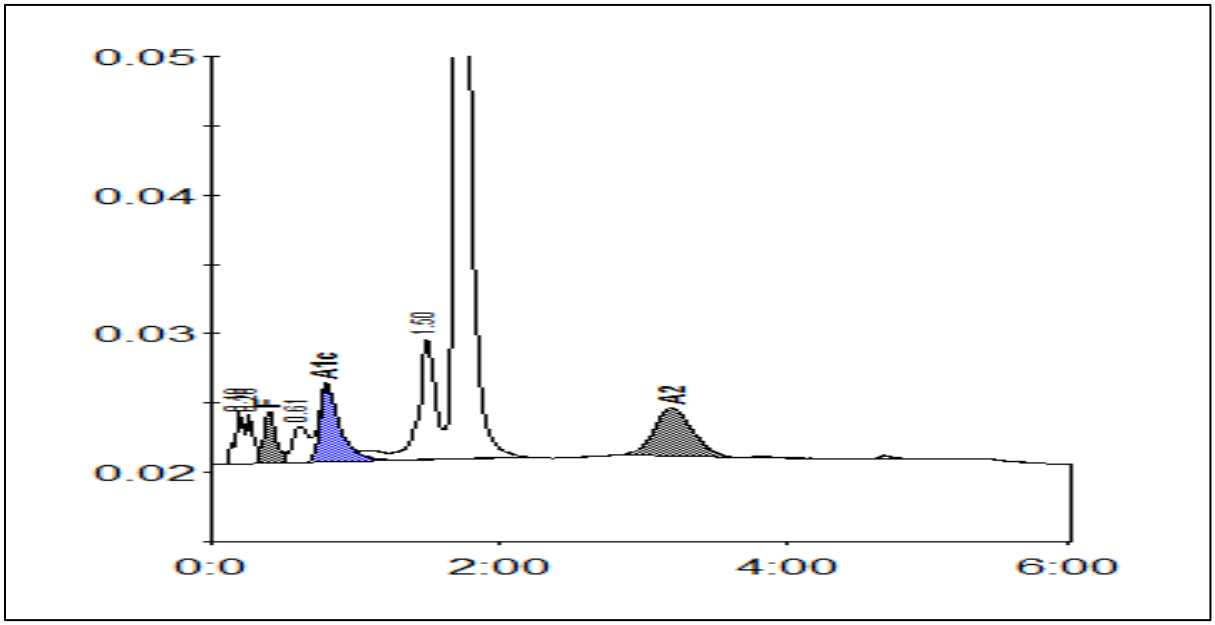
Picture 6: Peripheral blood Smear showing Normocytic normochromic red blood picture along with sickle cells in Sickle cell homozygous (Giemsa 400X)



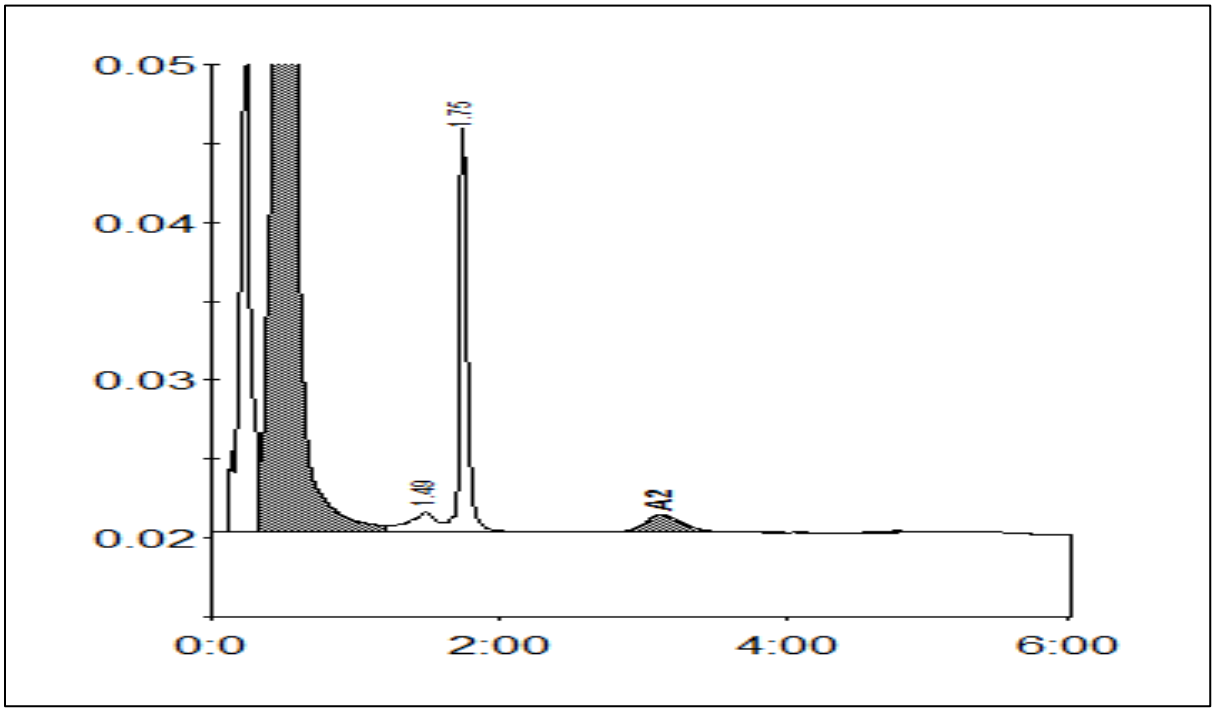
Picture 7: Peripheral Blood smear showing mild degree of anisocytosis with microcytic hypochromic RBC picture and occasional sickle cells in Compound heterozygous for sickle cell and β-thalassemia (Giemsa 400X)



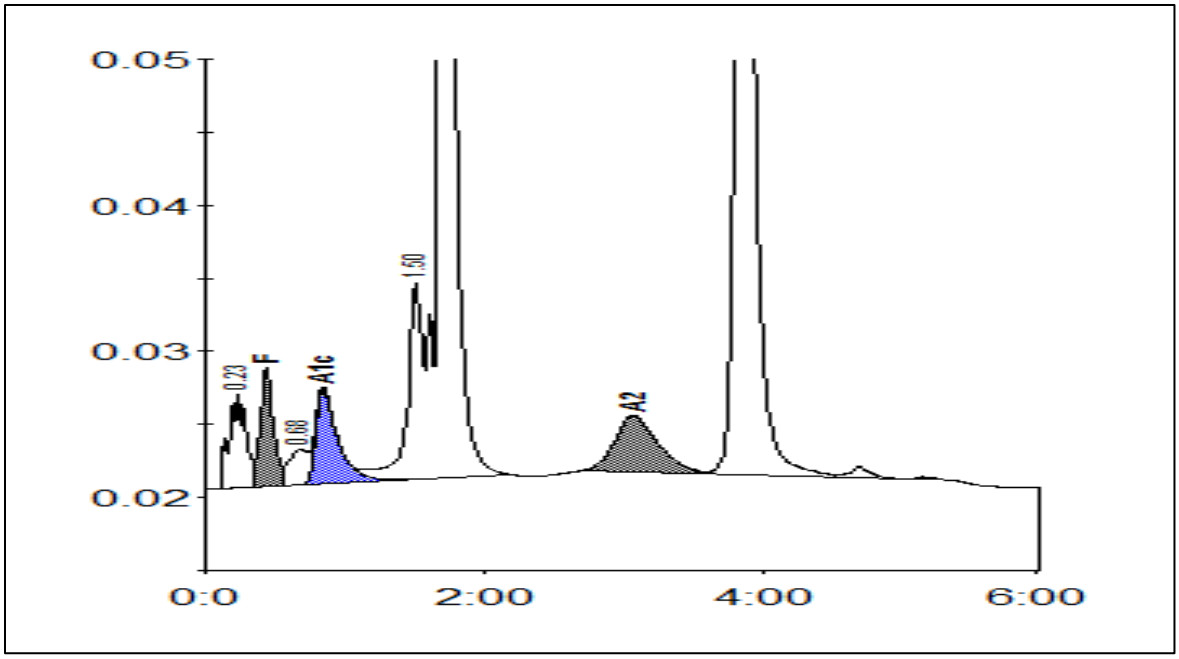
Picture 8: HPLC chromatogram showing a) HbF-0.5%, b) HbA-97.2% and HbA2-2.3% in normal patient



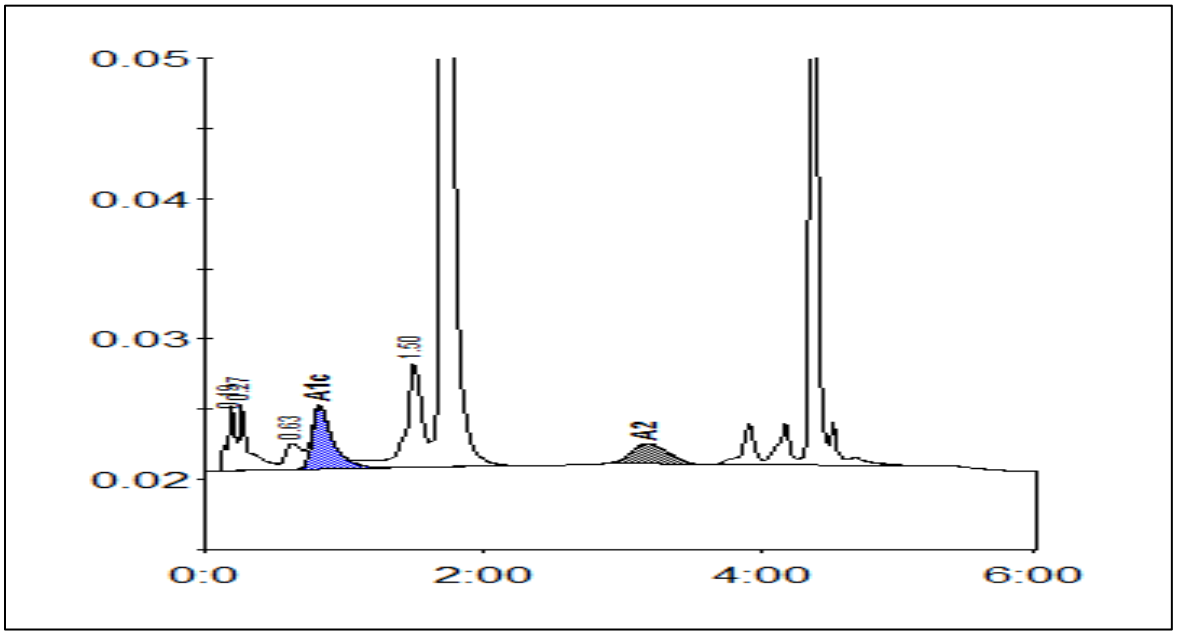
Picture 9: HPLC chromatogram showing a) HbF-1.5%, b) HbA2-6% and c) HbA-92.5% in β-thalassemia heterozygous



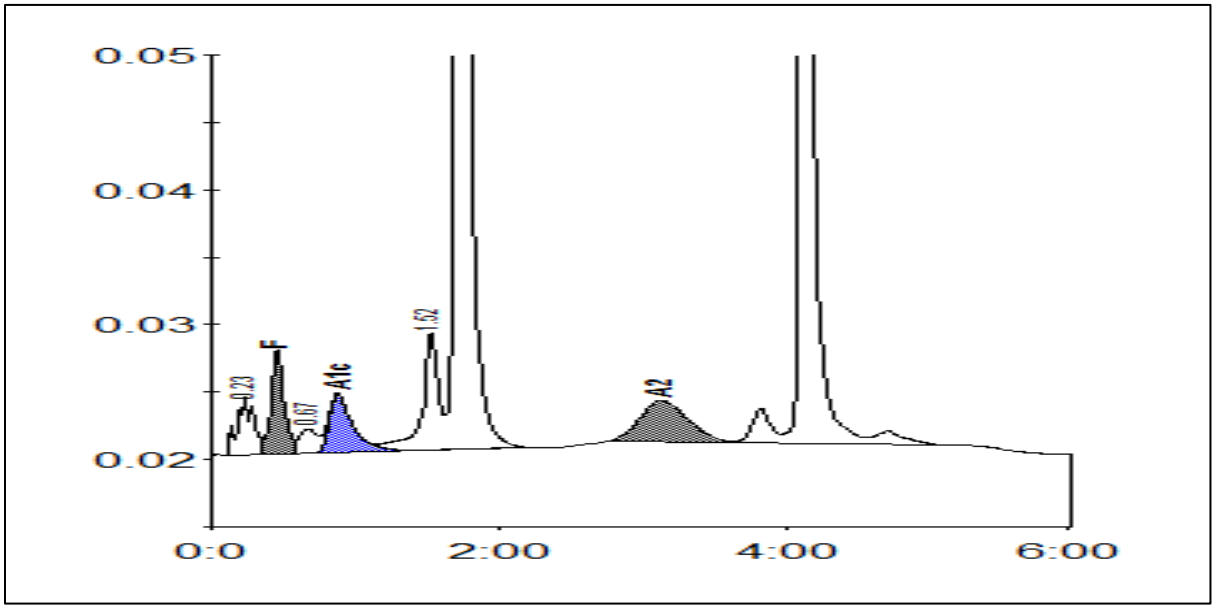
Picture 10: HPLC chromatogram showing a) HbF-75.5%, b) HbA2-2.2% and c) HbA-22.1% in β-Thalassemia major



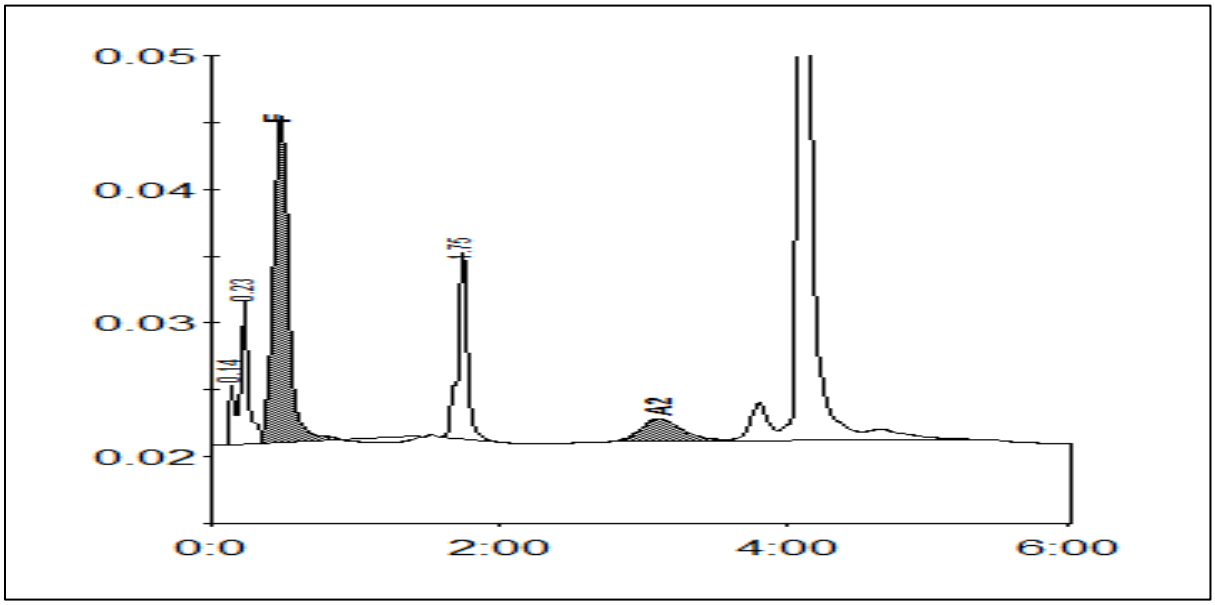
Picture 11: HPLC chromatogram showing a) HbD-28.1% at RT-3.8 minutes b) HbA-68.9%, c) HbA2-2.1%, d) HbF-0.9% in HbD Punjab heterozygous.



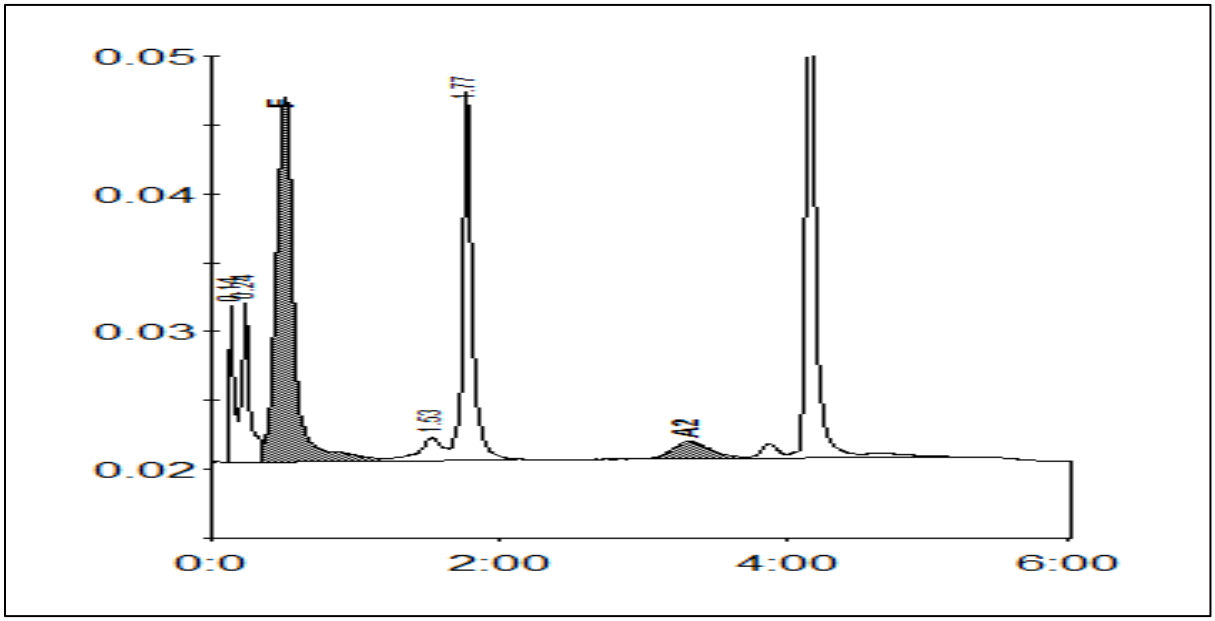
Picture 12: HPLC chromatogram showing a) HbQ-16.9% at RT- 4.37 minutes, b) HbA-80.4%, HbA2-2.7%, HbF-0.2% in HbQ India heterozygous



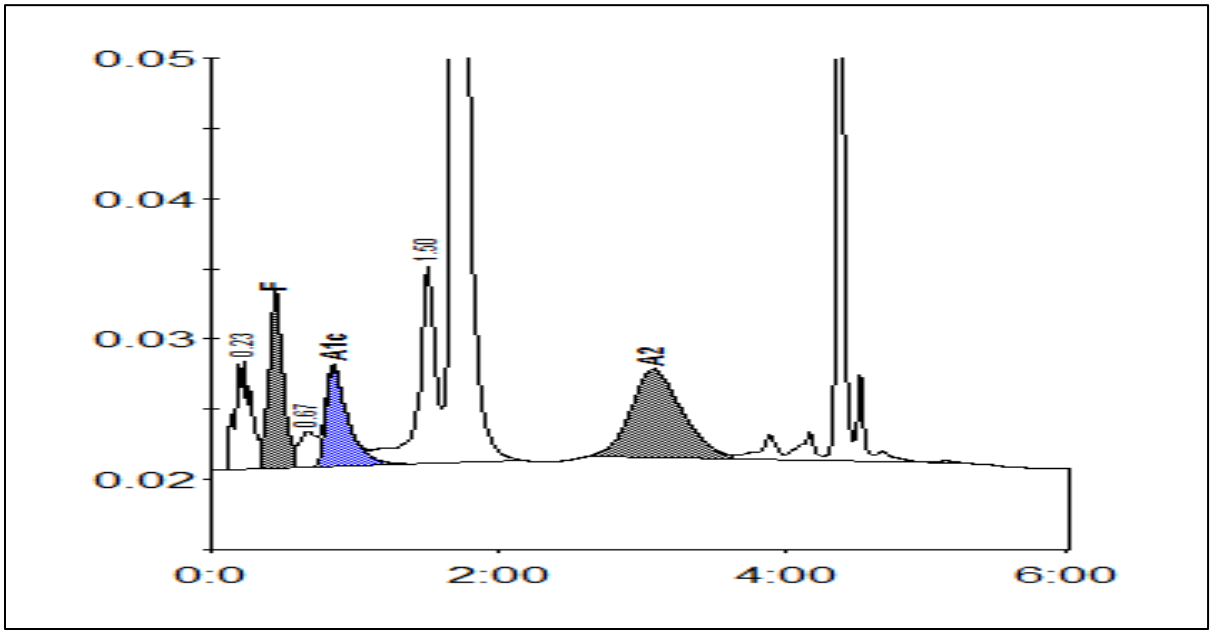
Picture 13: HPLC chromatogram showing a) HbS-31.1% at RT-4.12 minutes b) HbA2-3.1%, c) HbA-64.2%, d) HbF-1.6% in Sickle cell heterozygous



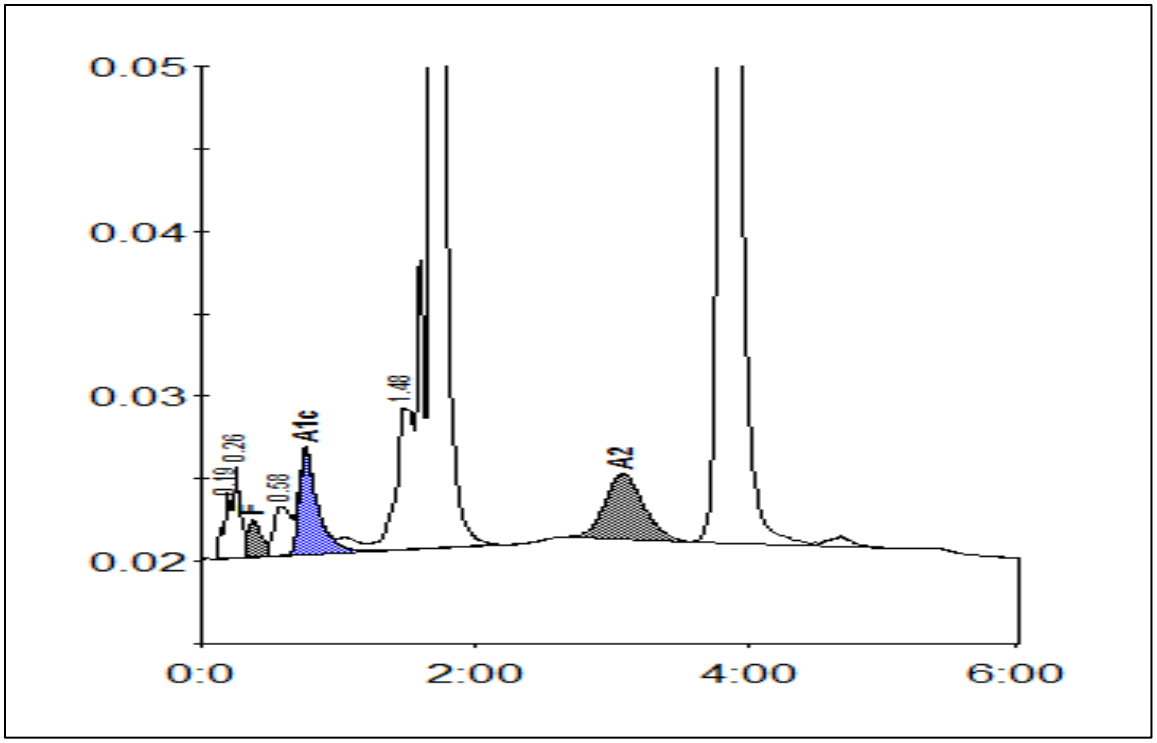
Picture 14: HPLC chromatogram showing a) HbS-66% at RT- 4.11 minutes, b) HbA-5.3%, c) HbA2-1.6%, d) HbF-24.5% in Sickle cell homozygous



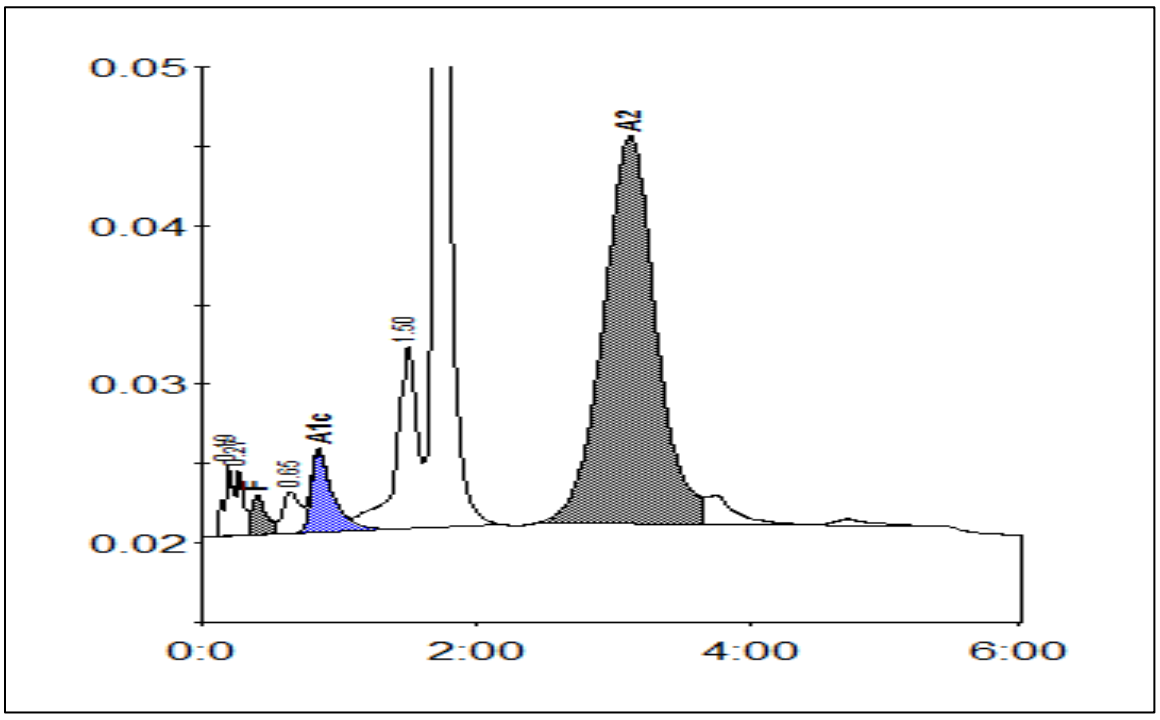
Picture 15: HPLC chromatogram showing a) HbS-34.9% at RT- 4.12 minutes, b) HbA2-4.0%, c) HbF-35.4%, d) HbA-18.3% in Compound heterozygote for Sickle cell and β-thalassemia



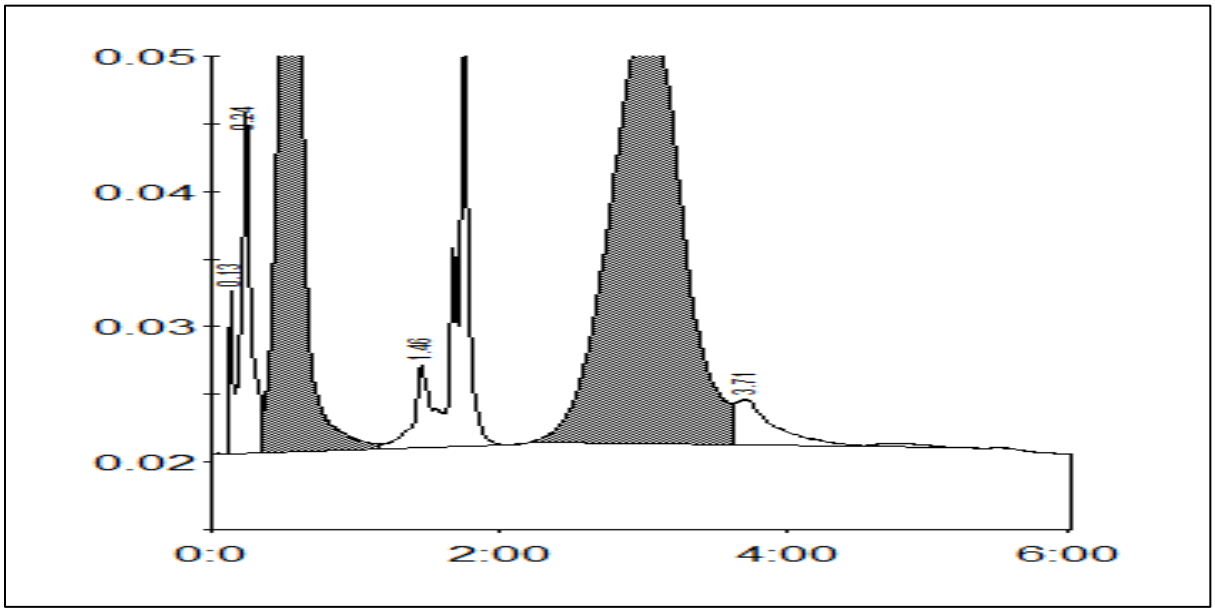
Picture 16: HPLC chromatogram showing a) HbQ-7.8% at RT- 4.35 minutes, b) HbA-82.6%, c) HbA2-6.7%, d) HbF-2.9% in Compound heterozygote for HbQ India and β-thalassemia.



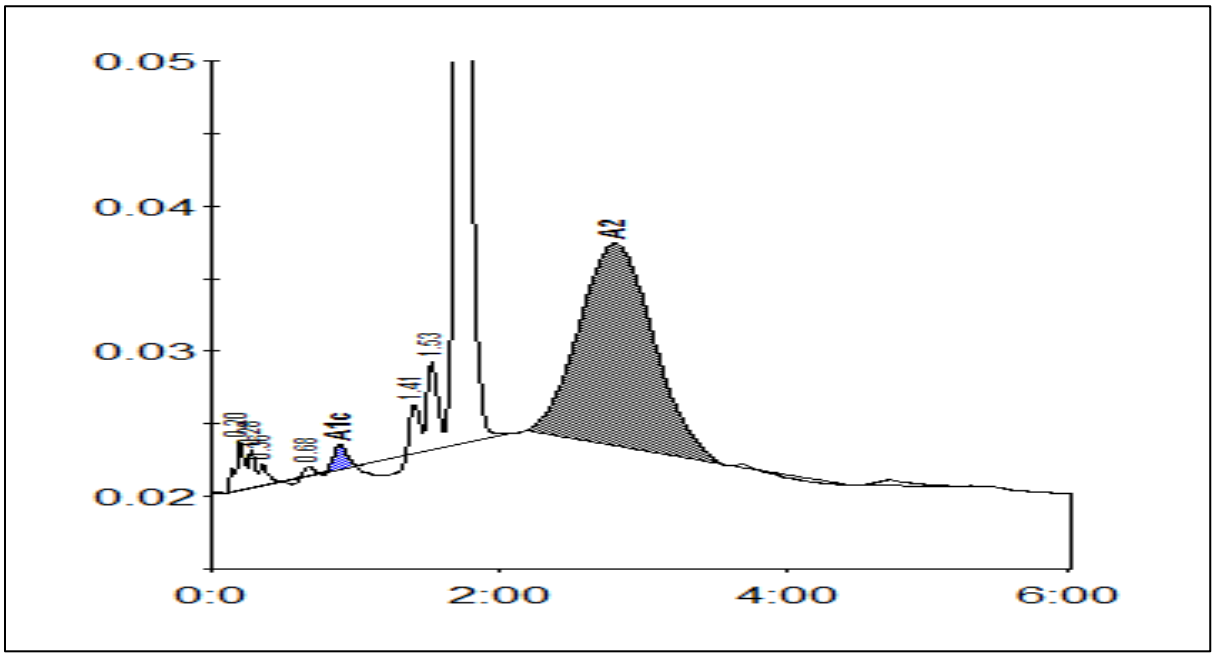
Picture 17: HPLC chromatogram showing a) HbD-78.5% at RT-3.8 minutes, b) HbA2-4.5%, HbA-7%, HbF-3.9% in Compound heterozygote for HbD Punjab and β-thalassemia



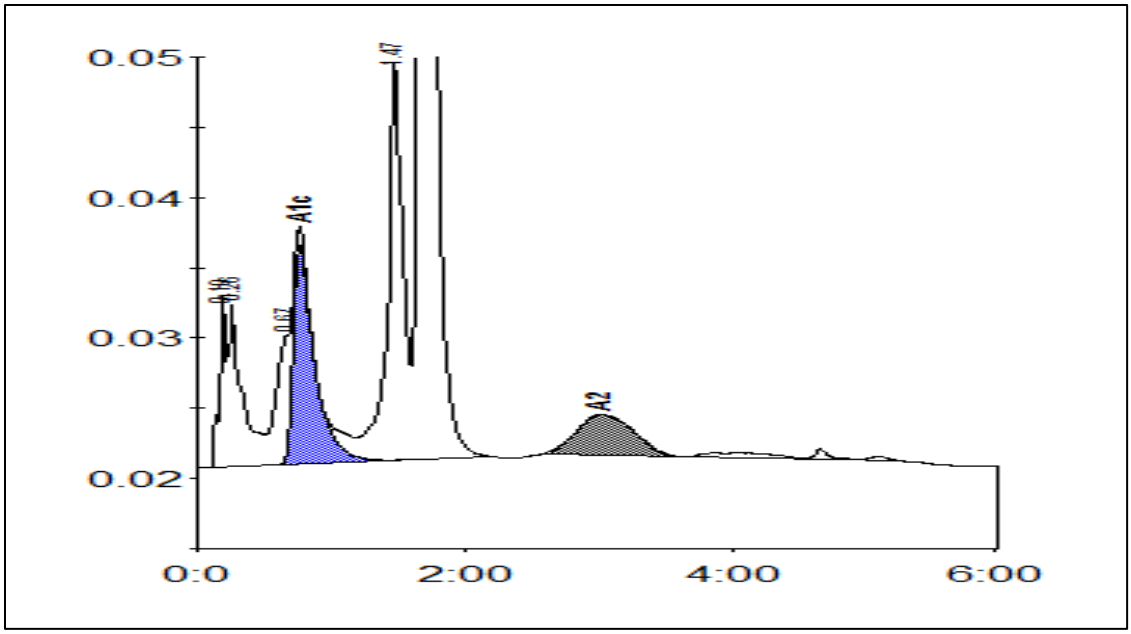
Picture 18: HPLC chromatogram showing a) HbA2-32.8%, b) HbA-66.5% c) HbF-0.7% in HbE heterozygous



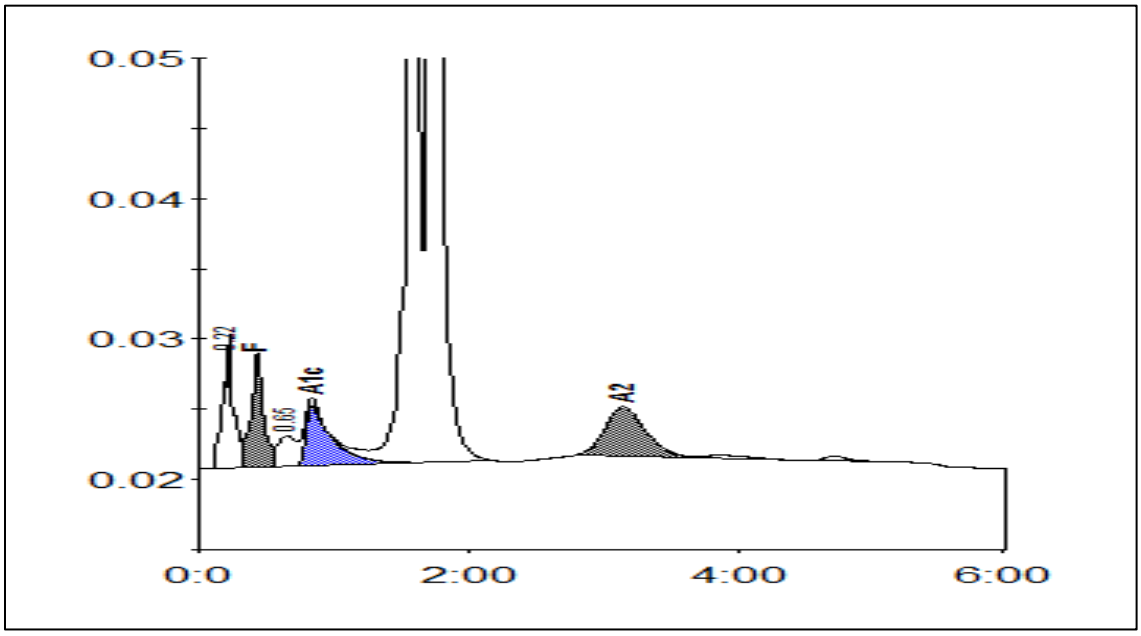
Picture 19: HPLC chromatogram showing a) HbA2-61.1%, b) HbA-7.2-%, c) HbF-31.7% in Compound heterozygous for HbE with β-thalassemia



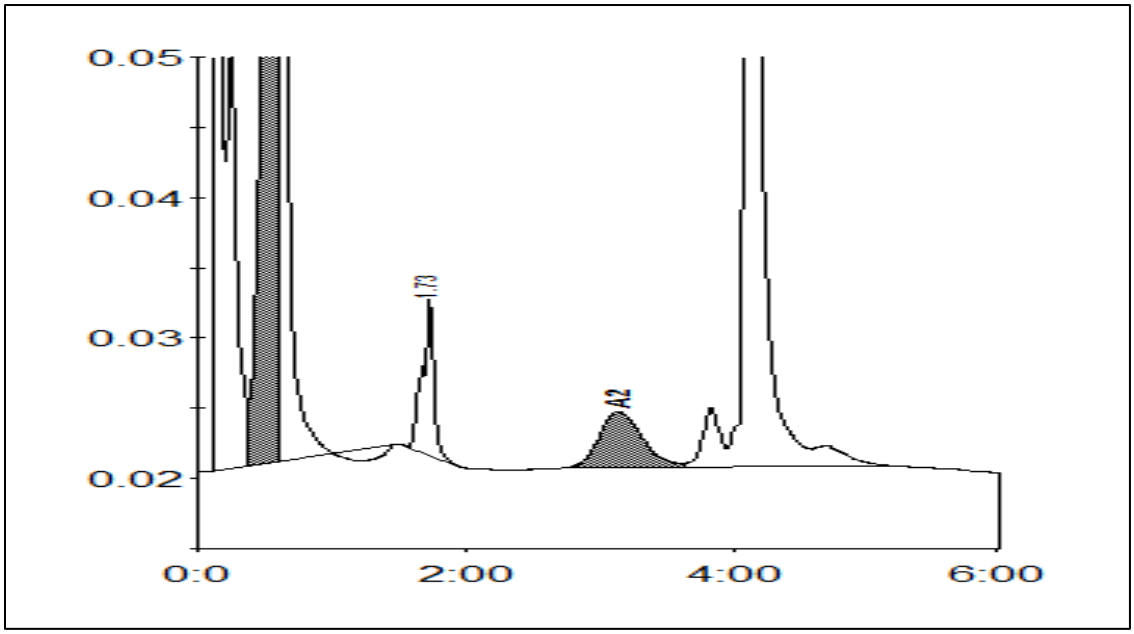
Picture 20: HPLC chromatogram showing a) HbA2-40.9%, b) HbA-59.1%, c) HbF-0.2% in HbD Iran heterozygous



Picture 21: HPLC chromatogram showing a) P3-7.4% at RT-1.47 minutes a) HbA2-3.3%, b) HbA-89.3%, c) HbF-0.2% in HbJ Meerut



Picture 22: HPLC chromatogram showing a) P3-23.4% at RT-1.57 minutes, b) HbA2- 4.4%, c) HbA-70.1%, HbF- 2.1% in Compound heterozygote for HbJ and β-thalassemia.



Picture 23: HPLC chromatogram showing a) HbS-50.6% at RT-4.11minutes, b) HbF-29.1%, c) HbA2-3.2%, HbA-2.2% in Compound heterozygote for HbS and Delta β-thalassemia

S.NO	MRD NO.	IMMUNOPATH NO.	AGE	SEX	PLACE OF BIRTH	CASTE/RELIGION	PALLOR	BLOOD TRANSFUSION RECEIVED	JAUNDICE	LIVER SPAN	SPLEEN SPAN	FAMILY HISTORY	RBC	Hb	MCV	MCH	MCHC	RDW	MENTZER INDEX	MICROCYTES	NORMOCYTES	PENCIL CELLS	TARGET CELLS	HYPOCHROMIA	NORMOCHROMIA	HbF	HbA	HbA2	HbD	Hb S	IMPRESSION
1	187139	34/2018	22	F	LUDHIANA	SIKH	NO	NO	NO				4.84	13	82	26.9	32.8	14.2	16.94	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96	4			MILDLY RAISED HbA2 LEVELS NOTED
2	413371	35/2018	23	F	LUDHIANA	HINDU	NO	NO	NO				4.7	14.2	88.9	29.6	33.3	14.6	18.59	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.3	3.7			BORDERLINE HbA2 LEVELS SEEN
3	415962	69/2018	24	F	KHANNA	HINDU	NO	NO	NO				4.81	11.1	70.7	23.1	32.6	17	14.69	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT		77.5	2.2			HBQ INDIA HETEROZYGOUS
4	414444	71/2018	1	M	PHILLAUR	SIKH	NO	NO	NO				3.13	4.1	52.9	13	24.6	35.7	16.9	ABSENT	ABSENT	ABSENT	ABSENT	ABSENT	ABSENT	0.9	68.9	2.1	28.1		HbD PUNJAB HETEROZYGOUS
5	415727	73/2018	33	F	LUDHIANA	HINDU	NO	NO	NO				3.95	12	90.8	30.3	33.4	14.1	22.98	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	95.7	3.6			BORDERLINE HbA2 LEVELS SEEN
6	3617	114/2018	38	F	LUDHIANA	PUNJABI	NO	NO	NO				4.53	13	84.6	28.8	34	13.2	18.67	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	95.6	3.7			BORDERLINE HbA2 LEVELS SEEN
7	414475	120/2018	1	M	JALANDHAR	HINDU	NO	NO	NO				4.89	7.1	52.1	14.6	27.9	27.8	10.65	PRESENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	1.1	67.2	1.7	30		HbD PUNJAB HETEROZYGOUS
8	417797	121/2018	31	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.11	9.6	60	18.8	31.3	16.6	11.74	PRESENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	1.5	92.5	6			BETA THALASSEMIA TRAIT
9	410911	210/2018	30	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.58	10.7	61.5	19.2	31.2	15.8	11.02	PRESENT	PRESENT	ABSENT	PRESENT	PRESENT	ABSENT	0.7	92.7	6.6			BETA THALASSEMIA TRAIT
10	403860	211/2018	30	F	LUDHIANA	HINDU	NO	NO	NO				3.79	10.8	86.9	28.4	32.7	14.1	22.92	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	95.7	3.6			MILDLY RAISED HbA2 LEVELS NOTED
11	420461	216/2018	38	F	LUDHIANA	HINDU	NO	NO	NO				4.51	12.3	82	27.1	33.1	13.2	18.18	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.3	3.7			MILDLY RAISED HbA2 LEVELS NOTED
12	421224	217/2018	29	F	SANGRUR	SIKH	NO	NO	NO				4.75	13	86.4	27.3	31.68	17.4	18.18	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.3	3.7			MILDLY RAISED HbA2 LEVELS NOTED
13	8199	260/2018	31	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.79	12.5	68.4	21.6	31.6	16.1	11.81	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	1	95.3	5.6			BETA THALASSEMIA TRAIT
14	240195	298/2018	66	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.62	10.7	61.9	19	30.8	17.2	11.01	PRESENT	PRESENT	ABSENT	PRESENT	PRESENT	ABSENT	2.3	91.9	5.8			BETA THALASSEMIA TRAIT
15	420456	307/2018	42	M	LUDHIANA	SIKH	NO	NO	NO				4.73	15.5	98.6	32.7	33.2	14.7	20.84	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	95.5	3.8			MILDLY RAISED HbA2 LEVELS NOTED
16	427689	398/2018	28	F	LUDHIANA	HINDU	NO	NO	NO				4.84	14.3	91.4	29.5	32.2	16.1	18.88	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.4	3.6			BORDERLINE HbA2 LEVELS SEEN
17	429030	401/2018	28	F	LUDHIANA	SIKH	NO	NO	NO			PRESENT	6.76	12.1	59.3	17.9	30.2	17.2	8.77	PRESENT	PRESENT	ABSENT	PRESENT	PRESENT	ABSENT	1.1	92.3	6.6			BETA THALASSEMIA TRAIT
18	427325	402/2018	24	F	LUDHIANA	HINDU	NO	NO	NO				3.99	12.1	91.4	30.4	33.3	14	22.9	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.1	3.9			BORDERLINE HbA2 LEVELS SEEN
19	3416	403/2018	15	F	LUDHIANA	HINDU	NO	NO	NO				4.8	12.6	87.5	26.2	32.2	17.3	16.97	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.4	3.6			BORDERLINE HbA2 LEVELS SEEN
20	408748	404/2018	29	F	LUDHIANA	SIKH	NO	NO	NO				4.58	12.8	85.8	27.8	32.5	14	18.73	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	95.6	3.7			BORDERLINE HbA2 LEVELS SEEN
21	413375	405/2018	29	M	LUDHIANA	HINDU	NO	NO	NO				4.67	14.3	95.3	30.6	32.1	14.8	20.4	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		95.8	4.2			MILDLY RAISED HbA2 LEVELS NOTED
22	297376	406/2018	8	F	LUDHIANA	SIKH	NO	NO	NO			PRESENT	5.5	10.1	58.7	18.4	31.3	18.5	10.67	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	6.9	87	6.1			BETA THALASSEMIA TRAIT
23	382024	407/2018	28	F	NEW DELHI	HINDU	NO	NO	NO				3.8	11.8	94.7	31	32.8	14.1	24.92	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.4	3.6			BORDERLINE HbA2 LEVELS SEEN
24	427575	451/2018	7	M	LUDHIANA	HINDU	NO	NO	NO			PRESENT	6.06	11.2	59.7	18.4	30.9	15.7	2.59	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	1	95.5	6.5			BETA THALASSEMIA TRAIT
25	356522	454/2018	31	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.42	12.1	70.6	22.4	31.7	16.4	13.02	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	4.9	88.1	7			BETA THALASSEMIA TRAIT
26	430731	460/2018	34	F	LUDHIANA	HINDU	YES	NO	NO				3.56	9.8	84.5	27.7	32.7	13.8	23.73	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	0.7	95.5	3.8			BORDERLINE HbA2 LEVELS SEEN
27	7123	461/2018	28	F	LUDHIANA	SIKH	NO	NO	NO				4.5	11.5	77.4	25.6	33	17.2	17.2	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	PRESENT		96.3	3.7			BORDERLINE HbA2 LEVELS SEEN
28	428753	480/2018	8	F	LUDHIANA	SIKH	YES	NO	NO	INCREASED	INCREASED	PRESENT	3.36	8	76.7	23.9	31.1	31.5	22.82	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	75.5	22.1	2.4			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
29	431545	483/2018	26	F	LUDHIANA	HINDU	NO	NO	NO				4.07	11.9	87.3	29.2	31.5	11.6	21.44	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	95.3	3.7			BORDERLINE HbA2 LEVELS SEEN
30	431759	489/2018	29	F	LUDHIANA	HINDU	NO	NO	NO			PRESENT	5.61	15.9	83.8	28.3	33.7	14.6	14.93	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	58.2	4	37.1		SUGGESTIVE OF COMPOUND HETEROZYGOUS FOR HbD PUNJAB & BETA THALASSEMIA
31	8230	493/2018	27	M	LUDHIANA	SIKH	NO	NO	NO				5.2	14.8	90.6	28.4	31.4	13.2	17.42	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.1	3.9			BORDERLINE HbA2 LEVELS SEEN
32	429260	494/2018	32	M	LUDHIANA	HINDU	NO	NO	NO				5.27	14.9	84.6	28.3	33.5	14	16.05	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.3	3.7			BORDERLINE HbA2 LEVELS SEEN
33	352263	570/2018	5	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.11	9.9	61.2	19.4	31.8	19.8	11.97	PRESENT	PRESENT	ABSENT	ABSENT	ABSENT	ABSENT	16	78.3	5.7			BETA THALASSEMIA TRAIT
34	356499	632/2018	36	M	JAGRAON	HINDU	NO	NO	NO				4.71	13.9	89.4	29.4	32.9	14.1	18.98	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.2	3.8			BORDERLINE HbA2 LEVELS SEEN
35	439253	675/2018	27	F	LUDHIANA	SIKH	NO	NO	NO				4.32	10.9	78.7	25.1	32	25.3	18.21	ABSENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT		75.5				SUGGESTIVE OF HbE TRAIT
36	424855	757/2018	26	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.76	10.1	55.7	17.5	31.5	17.5	9.67	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	0.8	92.8	6.4			BETA THALASSEMIA TRAIT
37	442560	761/2018	28	F	MOGA	SIKH	NO	NO	NO				5.09	13.2	78.6	26	33	15.1	15.44	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT		96.4	3.6			BORDERLINE HbA2 LEVELS SEEN
38	202728	762/2018	34	F	LUDHIANA	SIKH	NO	NO	NO				4.86	13.3	86.6	27.4	31.6	14.1	17.81	ABSENT	PRESENT	ABSENT	ABSENT	ABSENT	PRESENT	0.7	95.4	3.9			BORDERLINE HbA2 LEVELS SEEN
39	446203	861/2018	26	F	LUDHIANA	HINDU	NO	NO	NO				4.45	12.5	84.6	28	33.1	13	19.01	ABSENT	PRESENT	ABSENT	ABSENT		PRESENT	0.9	95.4	3.7			BORDERLINE HbA2 LEVELS SEEN
40	418221	865/2018	31	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.16	12.5	63.5	20.3	31.9	16.1	10.3	ABSENT	ABSENT	ABSENT	ABSENT		ABSENT	1.2	93.1	5.7			BETA THALASSEMIA TRAIT
41	345490	889/2018	33	F	JALANDHAR	HINDU	YES	NO	NO		INCREASED	PRESENT	4.88	10	64.5	20.6	31.9	15.9	13.21			ABSENT	ABSENT		ABSENT	2.9	82.6	6.7			COMPOUND HETEROZYGOUS HbQ AND BETA THALASSEMIA
42	449194	892/2018	34	M	LUDHIANA	HINDU	NO	NO	NO			PRESENT	5.7	13.2	73.1	23.1	31.6	17.3	12.82	PRESENT		ABSENT	ABSENT	PRESENT	ABSENT	0.7	91.9	7.4			BETA THALASSEMIA TRAIT
43	413375	893/2018	29	M	LUDHIANA	HINDU	NO	NO	NO				4.63	14.1	93.4	30.5	32.7	14.6	20.17		PRESENT	ABSENT	ABSENT		PRESENT		96.2	3.8			BORDERLINE HbA2 LEVELS SEEN
44	449071	951/2018	8	F	LUDHIANA	SIKH	YES	NO	NO				4.05	10.4	77.1	25.6	33.2	14	19.03		PRESENT	ABSENT	ABSENT	PRESENT	ABSENT		96.3	3.7			BORDERLINE HbA2 LEVELS SEEN
45	427050	953/2018	27	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.62	10.6	59.3	18.9	32	17.7	10.55	PRESENT		ABSENT	ABSENT	PRESENT	ABSENT	2.3	92.2	5.5			BETA THALASSEMIA TRAIT
46	450810	958/2018	20	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.23	11.6	60.1	18.6	30.9	16.8	9.64	PRESENT	PRESENT	ABSENT	ABSENT	PRESENT	ABSENT	1	95.6	6.4			BETA THALASSEMIA TRAIT
47	451779	959/2018	1	F	LUDHIANA	SIKH	NO	NO	NO				4.82	11.8	78	24.4	31.3	15.5	16.18		PRESENT	ABSENT	ABSENT		PRESENT	1.6	68.6	3.3	26.3		HbD PUNJAB HETEROZYGOUS
48	451227	961/2018	32	F	JALANDHAR	HINDU	NO	NO	NO				3.82	12.1	94.4	31.6	33.5	14.8	24.71		PRESENT	ABSENT	ABSENT		PRESENT	0.7	95	3.6			BORDERLINE HbA2 LEVELS SEEN
49	450136	962/2018	32	F	LUDHIANA	HINDU	NO	NO	NO				3.99	12.2	91.8	30.5	33.2	14.2	23		PRESENT	ABSENT	ABSENT		PRESENT	0.7	95.6	3.7			BORDERLINE HbA2 LEVELS SEEN
50	456734	1086/2018	25	F	LUDHIANA	SIKH	NO	NO	NO				4.14	12.3	90.9	29.7	32.7	16	21.95		PRESENT				PRESENT	0.7	61.3	2.5	35.5		HbD PUNJAB HETEROZYGOUS
51	95927	1087/2018	27	F	LUDHIANA	HINDU	NO	NO	NO				4.47	13.4	89.5	29.9	33.4	14	20.02		PRESENT				PRESENT	0.7	95.6	3.7			BORDERLINE HbA2 LEVELS SEEN
52	169834	1128/2018	61	F	JALANDHAR	SIKH	YES	NO	NO				3.41	8.8	77.3	25.7	33.2	15.8	22.66	PRESENT	PRESENT				PRESENT		89.3	3.3			HB J MEERUT
53	458358	1129/2018	23	F	LUDHIANA	HINDU	YES	NO	NO				3.65	10.2	85.7	27.9	32.5	14.4	23.47		PRESENT				PRESENT	0.7	95.5	3.8			BORDERLINE Hb2 LEVELS SEEN
54	442527	1133/2018	23	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	3.8	12.2	95.1	31.9	33.5	13.5	24.76		PRESENT				PRESENT	6	90.3	3.7			SUGGESTIVE OF BETA THALASSEMIA TRAIT
55	454345	1134/2018	31	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.2	12.5	64.7	20.1	31.1	14.9	10.43		PRESENT			PRESENT	ABSENT	1	92.9	6.1			BETA THALASSEMIA TRAIT
56	373485	1135/2018	35	F	JAMMU	HINDU	NO	NO	NO				4.2	12.6	89.3	30	33.5	14.1	21.26		PRESENT				PRESENT	0.7	57.3	3.5	38.5		HbD PUNJAB HETEROZYGOUS
57	457861	1168/2018	58	M		SIKH	YES	NO	NO			ABSENT	4.32	8.6	58	19.8	34.1	20.1	13.42	PRESENT	PRESENT				ABSENT	1	94.1	4.9			BETA THALASSEMIA TRAIT
58	35508	1232/2018	35	M	LUDHIANA	HINDU	YES	NO	NO		INCREASED		6.52	11.2	54.8	17.2	31.5	24.1	8.4	PRESENT				PRESENT	ABSENT	4.2	6	6.1	83.7		COMPOUND HETEROZYGOUS HbD PUNJAB AND BETA THALASSEMIA
59	345130	1235/2018	25	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.48	13.2	63.8	20.3	31.9	15.1	9.84	PRESENT				PRESENT	ABSENT	0.7	93.2	6.1			BETA THALASSEMIA TRAIT
60	257896	1236/2018	20	F	LUDHIANA	HINDU	NO	NO	NO				4.47	13.7	95.4	30.7	32.2	14.6	21.34		PRESENT				PRESENT	0.7	95.4	3.9			BORDERLINE HbA2 LEVELS SEEN
61	464077	1239/2018	50	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.95	9.5	61.8	19.1	31	16.4	12.48	PRESENT				PRESENT	ABSENT	0.7	92.6	6.7			BETA THALASSEMIA TRAIT
62	377727	1243/2018	21	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	4.34	12.1	85.8	28	32.7	14.4	19.76		PRESENT				PRESENT	0	95.8	4.2			SUGGESTIVE OF BETA THALASSEMIA TRAIT
63	461388	1246/2018	0.333333333	M	JALANDHAR	HINDU	YES	NO	NO	INCREASED	INCREASED		2.7	5.9	64.7	22	34	33	23.96		PRESENT				PRESENT	96.2	1.4	2.4			CLINICAL CORRELATION AND FAMILY SCREENING TO RULE OUT THALASSEMIA MAJOR
64	465564	1306/2018	27	F	LUDHIANA	SIKH	YES	NO	NO				4.56	13.1	85.1	28.6	33.7	13.3	18.66		PRESENT				PRESENT	0.7	55.5	3.4	40.4		HbD PUNJAB HETEROZYGOUS
65	326308	1350/2018	30	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.17	12	61.6	19.5	31.7	19.5	9.98	PRESENT				PRESENT	ABSENT	1.9	92.7	5.4			BETA THALASSEMIA TRAIT
66	473064	1486/2018	31	F	LUDHIANA	SIKH	NO	NO	NO			PRESENT	5.5	11.4	65.3	20.7	31.7	15.8	11.87	PRESENT	PRESENT				PRESENT	0.7	93	6.3			BETA THALASSEMIA TRAIT
67	476002	1541/2018	31	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.17	13	67.1	21	31.4	17	10.87	PRESENT	PRESENT				ABSENT	1.8	90.7	7.5			BETA THALASSEMIA TRAIT
68	483053	1972/2018	1	F	FAZILKA	SIKH	NO	NO	NO	INCREASED	INCREASED	PRESENT	3.98	9.7	72.7	24.3	33.4	26.7	18.26	PRESENT		PRESENT		PRESENT	ABSENT	62	36.2	1.8			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
69	443582	2039/2018	21	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.13	8.9	67.2	21.4	31.9	20.7	16.27	PRESENT	PRESENT			PRESENT	ABSENT	0.7	92.6	6.7			BETA THALASSEMIA TRAIT
70	472155	2040/2018	27	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.7	11.1	62.6	19.6	31.3	16.6	10.98	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93	6.3			BETA THALASSEMIA TRAIT
71	387112	2042/2018	33	F	LUDHIANA	SIKH	NO	NO	NO				4.11	12	86.1	29	33.7	14.7	20.94		PRESENT				PRESENT		80.4	2.7			HbQ INDIA HETEROZYGOUS
72	494364	2047/2018	28	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.11	8.9	57.8	17.5	30.2	19.6	11.31	PRESENT	PRESENT		PRESENT	PRESENT	ABSENT	0.7	92.8	6.5			BETA THALASSEMIA TRAIT
73	499448	2197/2018	27	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	4.15	13	91.3	31.3	34.3	14.4	22		PRESENT				PRESENT	0.7	95.3	4			SUGGESTIVE OF BETA THALASSEMIA TRAIT
74	500868	2205/2018	26	F	KAPURTHALA	HINDU	YES	NO	NO			ABSENT	4.14	8.9	69.6	21.4	30.8	20	16.81	PRESENT	PRESENT			PRESENT	ABSENT	7	87.6	5.4			BETA THALASSEMIA TRAIT
75	228599	2206/2018	20	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.78	10	56.2	17.3	30.8	18.6	9.72	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.9	5.4			BETA THALASSEMIA TRAIT
76	486138	2210/2018	50	M	FATEHGARH SAHIB	HINDU	YES	NO	NO	PRESENT	ABSENT	ABSENT	5.65	11.2	62.2	19.8	31.8	19.8	11.18	PRESENT	PRESENT			PRESENT	ABSENT	1.5	92.8	5.7			BETA THALASSEMIA TRAIT
77	502804	2239/2018	20	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.21	9.3	58.5	17.8	30.4	26.5	11.22		PRESENT			PRESENT	ABSENT	1.2	92.9	5.9			BETA THALASSEMIA TRAIT

S.NO	MRD NO.	IMMUNOPATH NO.	AGE	SEX	PLACE OF BIRTH	CASTE/RELIGION	PALLOR	BLOOD TRANSFUSION RECEIVED	JAUNDICE	LIVER SPAN	SPLEEN SPAN	FAMILY HISTORY	RBC	Hb	MCV	MCH	MCHC	RDW	MENTZER INDEX	MICROCYTES	NORMOCYTES	PENCIL CELLS	TARGET CELLS	HYPOCHROMIA	NORMOCHROMIA	HbF	HbA	HbA2	HbD	Hb S	IMPRESSION
78	493161	2242/2018	59	F	KANGRA	HINDU	NO	YES	NO			ABSENT	3.52	6.4	57.5	17.9	31.2	18.7	16.06	PRESENT	PRESENT		PRESENT	PRESENT	ABSENT	0.7	86.6	5.6			BETA THALASSEMIA TRAIT
79	369640	2295/2018	29	F	LUDHIANA	HINDU	NO	NO	NO			PRESENT	5.21	10.7	64.9	20.6	31.8	15.7	12.45	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.6	5.7			BETA THALASSEMIA TRAIT
80	507277	2373/2018	29	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	6.47	12.6	65.1	19.5	30	16.9	10.06	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.4	5.9			BETA THALASSEMIA TRAIT
81	500719	2377/2018	63	M	SANGRUR		NO	NO	NO			ABSENT	5.87	11.9	65.4	20.2	30.9	17.7	11.14	PRESENT	PRESENT			PRESENT	ABSENT	1.1	93	5.9			BETA THALASSEMIA TRAIT
82	505757	2381/2018	64	F	JAMMU	SIKH	YES	NO	NO			ABSENT	3.06	6.9	71.8	23.4	31.2	26.7	23.46	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.8	5.5			BETA THALASSEMIA TRAIT
83	508052	2382/2018	33	F	JALANDHAR	SIKH	YES	NO	NO			ABSENT	4.26	8.5	63.3	20	31.6	19.1	14.85	PRESENT	PRESENT			PRESENT	ABSENT	1.8	91.9	6.3			BETA THALASSEMIA TRAIT
84	474747	2425/2018	23	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.37	10.4	61	19.4	31.8	16.6	11.35	PRESENT	PRESENT			PRESENT	ABSENT	1.5	92.5	6			BETA THALASSEMIA TRAIT
85	510365	2428/2018	40	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.47	11.3	66.3	20.6	31.1	15.9	12.12	PRESENT	PRESENT			PRESENT	ABSENT	1	93.5	5.5			BETA THALASSEMIA TRAIT
86	509882	2433/2018	20	M	LUDHIANA	SIKH	YES	NO	NO				5.16	15.9	94.3	30.9	32.8	12.8	18.27		PRESENT				PRESENT		96.2	3.8			BORDERLINE HbA2 LEVELS SEEN
87	252471	2491/2018	49	F	BHATINDA	HINDU	NO	NO	NO			ABSENT	5.08	9.7	61.5	19	30.9	17.4	12.1	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.8	5.5			BETA THALASSEMIA TRAIT
88	511343	2492/2018	28	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	3.87	9.8	77.9	25.2	25.3	15.3	20.12	PRESENT	PRESENT			PRESENT	ABSENT	2.4	90.6	7			BETA THALASSEMIA TRAIT
89	511298	2494/2018	40	M	LUDHIANA	HINDU	NO	NO	NO	INCREASED	INCREASED	PRESENT	4.57	10.6	72.2	23.1	32	16.2	15.79	PRESENT	PRESENT				ABSENT	1.2	92.8	6			BETA THALASSEMIA TRAIT
90	216564	2509/2018	29	F	LUDHIANA	SIKH	NO	NO	NO				4.44	13.8	90.9	31.2	34.3	15.5	20.47		PRESENT				PRESENT	0.7	57.6	2.9	38.8		HbD PUNJAB HETEROZYGOUS
91	509605	2675/2018	24	F	LUDHIANA	HINDU	NO	YES	NO			PRESENT	3.98	8.6	70.4	21.5	30.6	15.8	17.68	PRESENT	PRESENT			PRESENT	ABSENT	1.3	93.6	5.1			BETA THALASSEMIA TRAIT
92	517623	2685/2018	20	F	LUDHIANA	HINDU	YES	YES	NO				1.38	5	103.6	36.2	34.9	16.3	75.07		1				PRESENT	0.7	85.3	3	11		HbD PUNJAB HETEROZYGOUS
93	284254	2760/2018	28	F	LUDHIANA	JAINSM	YES	NO	NO			ABSENT	5.45	10.6	61.9	19.5	31.5	17.9	11.35	PRESENT	PRESENT			PRESENT	ABSENT	0.7	94.6	4.7			BETA THALASSEMIA TRAIT
94	192903	2762/2018	32	F	MOGA	SIKH	NO	NO	NO	INCREASED			3.76	11.3	88.8	30	33.8	14.1	23.61		PRESENT				PRESENT	0.7	58.8	3.1	37.4		HbD PUNJAB HETEROZYGOUS
95	352263	2770/2018	10	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.48	10.4	59.8	19	31.7	18.8	10.91	PRESENT	PRESENT			PRESENT	ABSENT	8.7	85.5	5.8			BETA THALASSEMIA TRAIT
96	297376	2771/2018	1	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.67	10.5	60.3	18.5	30.7	18.4	10.63	PRESENT	PRESENT			PRESENT	ABSENT	6.1	88.6	5.3			BETA THALASSEMIA TRAIT
97	523303	2835/2018	32	F	LUDHIANA	SIKH	YES	NO	NO				4.06	10.9	81.6	26.9	32.9	14.5	20.09		PRESENT				PRESENT	0.7	62.9	2.8	33.6		HbD PUNJAB HETEROZYGOUS
98	524166	2840/2018	40	M	LUDHIANA	HINDU	YES	NO	NO				5.32	13.7	77.2	25.8	33.4	16.6	14.51		PRESENT				PRESENT	0.7	66.5	32.8			SUGGESTIVE OF HbE HETEROZYGOUS
99	527172	2938/2018	29	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.3	11.7	83.4	27.2	32.6	14.4	19.39		PRESENT				PRESENT	0.7	95.2	4.1			SUGGESTIVE OF BETA THALASSEMIA TRAIT
100	500652	2994/2018	28	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	3.38	8.2	78.5	24.4	31	18.4	23.22		PRESENT				PRESENT	1.5	91.7	6.8			BETA THALASSEMIA TRAIT
101	533537	3103/2018	32	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.22	12.2	62.2	19.6	31.4	20.6	10	PRESENT		PRESENT		PRESENT	ABSENT	0.7	93.8	5.5			BETA THALASSEMIA TRAIT
102	533111	3106/2018	20	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.39	11.1	66.5	20.7	31.1	16.9	12.33	PRESENT	PRESENT			PRESENT	ABSENT	0.9	92.3	6.8			BETA THALASSEMIA TRAIT
103	116592	3107/2018	34	F	LUDHIANA	HINDU	NO	NO	NO				4.35	12.3	85.5	28.3	33.1	13.5	19.65		PRESENT				PRESENT	0.7	95.6	3.7			BORDERLINE HbA2 LEVELS SEEN
104	531458	3108/2018	72	M	MALERKOTLA (SANGRUR)	HINDU	YES	NO	NO	NORMAL	INCREASED		4.41	10.7	76.6	24.3	31.8	16.4	17.36	PRESENT	PRESENT			PRESENT	ABSENT		14.8	4.2	81		COMPOUND HETEROZYGOUS HbD PUNJAB AND BETA THALASSEMIA
105	355673	3113/2018	27	M	LUDHIANA	JAINSM	NO	NO	NO			ABSENT	5.35	11.2	65.1	20.9	32.1	15.1	12.16	PRESENT			PRESENT		ABSENT	0.7	92.89	6.4			BETA THALASSEMIA TRAIT
106	535480	3124/2018	38	F	HOSHIARPUR	HINDU	YES	NO	NO			ABSENT	4.85	10.1	65.2	20.9	32.1	16.7	13.44	PRESENT				PRESENT	ABSENT	1.1	93.4	5.5			BETA THALASSEMIA TRAIT
107	537073	3164/2018	29	F	BHATINDA	SIKH	NO	NO	NO			ABSENT	4.37	9.1	68	20.9	30.1	15.5	15.56	PRESENT				PRESENT	ABSENT	0.7	94.6	4.7			BETA THALASSEMIA TRAIT
108	536337	3167/2018	29	F	SANGRUR	SIKH	YES	NO	NO			ABSENT	4.5	10.7	73.8	23.7	32.1	23.5	16.4		PRESENT			PRESENT	ABSENT	0.7	94.7	4.6			BETA THALASSEMIA TRAIT
109	536356	3240/2018	22	F	JAMMU	HINDU	NO	NO	NO				1.77	7	120.2	39.6	32.9	31.1	67.9		PRESENT			PRESENT	ABSENT	0.8	95.5	3.7			BORDERLINE HbA2 LEVELS SEEN
110	538305	3241/2018	34	M	USHA RANI	HINDU	NO	NO	NO			ABSENT	6.55	12.8	64.5	19.6	30.4	17.6	9.84	PRESENT				PRESENT	ABSENT	0.7	93.4	5.9			BETA THALASSEMIA TRAIT
111	539243	3245/2018	32	F	LUDHIANA	SIKH	NO	NO	NO				3.99	8.3	68.6	20.8	30.3	18.7	17.19	PRESENT				PRESENT	ABSENT	0.7	81.9	2.1			HbQ INDIA HETEROZYGOUS
112	538966	3247/2018	31	F	LUDHIANA	SIKH	NO	NO	NO				3.68	11.3	89.5	30.8	34.4	14.1	24.32		PRESENT				PRESENT	0.7	57.5	2.7	39.1		HbD PUNJAB HETEROZYGOUS
113	540781	3294/2018	23	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.61	9	62.9	19.6	31.1	16.9	13.64	PRESENT				PRESENT	ABSENT	0.7	93.5	5.8			BETA THALASSEMIA TRAIT
114	542917	3343/2018	23	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	4.72	10.3	66.9	21.9	32.6	16.8	14.17	PRESENT	PRESENT			PRESENT	ABSENT	2.3	91.9	5.8			BETA THALASSEMIA TRAIT
115	543690	3404/2018	29	F	LUDHIANA	SIKH	NO	NO	NO		INCREASED	ABSENT	5.12	10	63	19.5	31	16.6	12.3	PRESENT				PRESENT	ABSENT	2.1	70.1	4.4			SUGGESTIVE OF COMPOUND HETEROZYGOUS FOR HbJ-VARIANT AND BETA THALASSEMIA TRAIT
116	537954	3451/2018	28	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	4.47	11.5	77.8	25.8	33.1	14.4	17.4		PRESENT				PRESENT	0.7	95.1	4.2			SUGGESTIVE OF BETA THALASSEMIA TRAIT
117	89187	3460/2018	19	M	AMRITSAR	HINDU	NO	NO	NO			ABSENT	4.31	11.9	84	27.5	32.7	16.6	19.48		PRESENT				PRESENT	17.1	8.4	3.5		71	SICKLE CELL HOMOZYGOUS (HbS)
118	549266	3464/2018	37	F	LUDHIANA	HINDU	NO	NO	NO				4.26	13.5	93.2	31.8	34.1	13.7	21.87		PRESENT				PRESENT		96.1	3.9			BORDERLINE HbA2 LEVELS SEEN
119	550858	3492/2018	27	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.35	9.7	59.8	18.1	30.2	19	11.17	PRESENT	PRESENT				PRESENT	0.7	94.6	4.7			BETA THALASSEMIA TRAIT
120	553874	3562/2018	11	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.3	11.1	58.2	17.7	30.4	16.7	9.23	PRESENT	PRESENT			PRESENT	ABSENT	0.8	93.6	5.6			BETA THALASSEMIA TRAIT
121	504105	3566/2018	25	M	FARIDKOT	HINDU	YES	NO	NO			ABSENT	4.95	9.4	60.7	19	31.2	15.5	12.26	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.3	6			BETA THALASSEMIA TRAIT
122	8023	3719/2018	6	M	MOGA	SIKH	YES	NO	NO			ABSENT	5.53	10.4	58.5	18.9	32.3	16.8	10.57	PRESENT	PRESENT	PRESENT		PRESENT	ABSENT	0.7	93.7	5.6			BETA THALASSEMIA TRAIT
123	563311	3787/2018	36	F	LUDHIANA	HINDU	YES	NO	NO				3.37	9	83.2	26.8	32.2	24.3	24.68	PRESENT	PRESENT			PRESENT	ABSENT	0.7	58.8	2.9	37.6		HbD PUNJAB HETEROZYGOUS
124	559234	3789/2018	16	F	LUDHIANA	JAINSM	NO	NO	NO			PRESENT	5.5	9.9	57.5	18	31.2	19.4	10.45	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.9	5.4			BETA THALASSEMIA TRAIT
125	8829	3831/2018	48	M	DEORIA	SIKH	YES	YES	YES			ABSENT	2.34	6.1	80.7	25.9	32	32.3	34.48	PRESENT	PRESENT			PRESENT	ABSENT	0.8	92.9	6.3			BETA THALASSEMIA TRAIT
126	552197	3834/2018	32	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.04	10.7	68.3	21.2	31.1	16.3	13.55	PRESENT	PRESENT			PRESENT	ABSENT	0.7	93.9	5.4			BETA THALASSEMIA TRAIT
127	557667	3835/2018	54	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.57	10.9	60.2	19.6	32.5	17.9	10.8	PRESENT	PRESENT			PRESENT	ABSENT	2.4	91.9	5.7			BETA THALASSEMIA TRAIT
128	568812	3894/2018	0.75	M	MOGA	SIKH	NO	YES	NO	INCREASED	INCREASED	PRESENT	1.01	3.3	74.8	32.9	44	37	74.05	PRESENT	PRESENT	PRESENT			ABSENT	82.3	13.9	3.8			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
129	557908	3896/2018	35	F	LUDHIANA	HINDU	NO	NO	NO			PRESENT	5.45	9.2	55.2	16.9	30.6	21.6	12.4	PRESENT	PRESENT			PRESENT	ABSENT	0.7	94.2	5.1			BETA THALASSEMIA TRAIT
130	569129	3897/2018	27	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.82	9.3	63.3	19.4	30.6	17.2	13.13	PRESENT	PRESENT			PRESENT	ABSENT	1.1	93.2	5.7			BETA THALASSEMIA TRAIT
131	559727	3898/2018	31	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.45	10.9	63.8	20.1	31.5	16.3	11.7	PRESENT				PRESENT	ABSENT	0.7	93.9	5.4			BETA THALASSEMIA TRAIT
132	48650	3977/2018	9	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.17	10	63	19.3	30.9	15.6	12.18	PRESENT	PRESENT			PRESENT	ABSENT	2.2	92.1	5.7			BETA THALASSEMIA TRAIT
133	570042	3992/2018	23	F	JALANDHAR	HINDU	YES	NO	NO			PRESENT	3.71	8.1	72	22	30.5	16.2	19.4	PRESENT	PRESENT			PRESENT	ABSENT	1.6	92.5	5.9			BETA THALASSEMIA TRAIT
134	574269	4079/2018	2	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.12	10.6	58	17.4	30	16.9	9.47	PRESENT	PRESENT	PRESENT			ABSENT	3.2	91.3	5.5			BETA THALASSEMIA TRAIT
135	542422	4080/2018	27	F	LUDHIANA	HINDU	NO	NO	NO			PRESENT	3.82	10.9	87	28.5	32.7	14.1	27.01		PRESENT				PRESENT	1.6	64.2	3.1		31.1	SICKLE CELL HETEROZYGOUS (HbS)
136	577122	4122/2018	25	F	LUDHIANA	HINDU	YES	NO	NO				5.04	9.3	63	18.5	29.5	17.1	12.5	PRESENT				PRESENT	ABSENT	1	92.3	6.7			BETA THALASSEMIA TRAIT
137	580567	4222/2018	23	M	HOSHIARPUR	SIKH	NO	NO	NO			ABSENT	7.36	14.1	64	19.2	29.8	15.7	8.69	PRESENT				PRESENT	ABSENT	0.7	93.8	5.5			BETA THALASSEMIA TRAIT
138	570165	4277/2018	5	M	SANGRUR	HINDU	YES	NO	NO				4.53	9.5	67	21.1	31.4	31.5	14.79	PRESENT				PRESENT	ABSENT	0	73.3	2.4	24.3		HbD PUNJAB HETEROZYGOUS
139	373485	4349/2018	35	F	JAMMU	HINDU	NO	NO	NO				4.08	12.3	92	30.2	32.9	14.3	22.54		PRESENT				PRESENT	0.7	60.1	2.5	36.7		HbD PUNJAB HETEROZYGOUS
140	574895	4468/2018	60	M	JAMMU	SIKH	YES	NO	NO			ABSENT	3.84	9.2	78	24	30.7	18.1	20.31	PRESENT	PRESENT			PRESENT	ABSENT	1.4	92.6	6			BETA THALASSEMIA TRAIT
141	590437	4470/2018	28	F	LUDHIANA	HINDU	NO	YES	NO			PRESENT	5	10	63.5	20	31.5	17.3	12.7	PRESENT	PRESENT			PRESENT	ABSENT	1.3	93.6	5.1			BETA THALASSEMIA TRAIT
142	583527	4474/2018	28	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.99	12.2	67	20.3	30.4	16.4	11.18	PRESENT	PRESENT			PRESENT	ABSENT	1.4	93.6	5			BETA THALASSEMIA TRAIT
143	589738	4480/2018	14	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	3.87	7.9	67	20.4	30.6	21.4	17.31	PRESENT	PRESENT	PRESENT		PRESENT	ABSENT	7.6	87.1	5.3			BETA THALASSEMIA TRAIT
144	592994	4505/2018	58	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.51	11.6	57.6	17.9	31.1	17.5	8.84	PRESENT				PRESENT	ABSENT	0	95.5	4.5			BETA THALASSEMIA TRAIT
145	537042	4514/2018	48	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.43	9.6	64.9	21.7	33.4	17.7	14.65	PRESENT	PRESENT			PRESENT	ABSENT	3.6	92.2	4.2			BETA THALASSEMIA TRAIT
146	594685	4608/2018	26	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.13	10.6	65.3	20.6	31.5	14.8	12.72	PRESENT				PRESENT	ABSENT	0.9	93.5	5.6			BETA THALASSEMIA TRAIT
147	594358	4609/2018	29	F	KATHUA	HINDU	NO	NO	NO			ABSENT	5.5	10.9	64.7	19.9	30.8	17.2	11.76		PRESENT			PRESENT	ABSENT	0.7	93.2	6.1			BETA THALASSEMIA TRAIT
148	485381	4615/2018	35	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	7.55	14.2	63	18.8	29.8	16	8.34		PRESENT			PRESENT	ABSENT	0.9	93.8	5.3			BETA THALASSEMIA TRAIT
149	596804	4660/2018	32	F	SBS NAGAR	SIKH	NO	NO	NO				4.19	10	71.6	23.9	33.4	18.7	17.08	PRESENT	PRESENT			PRESENT	ABSENT	0	65.8	1.7	32.5		HbD PUNJAB HETEROZYGOUS
150	598010	4752/2018	2	M	LUDHIANA	SIKH	NO	NO	NO	INCREASED	INCREASED	PRESENT	2	5.6	69	28	40.5	22.5	34.5	PRESENT	PRESENT				ABSENT	44	53.74	2.3			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
151	598288	4743/2018	25	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.6	10.3	62	18.1	29.4	16.1	10.91	PRESENT	PRESENT			PRESENT		0.7	94.6	4.7			BETA THALASSEMIA TRAIT
152	601716	4814/2018	26	F	FAZILKA	HINDU	NO	NO	NO	INCREASED	INCREASED	ABSENT	1.38	3.9	94	28.1	29.8	22.1	68.11	PRESENT	PRESENT			PRESENT		59.5	38.4	2.1			BETA THALASSEMIA HOMOZYGOUS
153	490641	4882/2018	25	F	HOSHIARPUR	HINDU	NO	NO	NO			ABSENT	5.3	10.7	66	20.2	30.8	14.7	12.45	PRESENT	PRESENT			PRESENT		2.1	93.7	4.2			BETA THALASSEMIA TRAIT
154	605561	4943/2018	24	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.2	8.3	64.1	19.8	30.8	25.1	15.26	PRESENT	PRESENT		PRESENT			0.7	94.9	4.4			SUGGESTIVE OF BETA THALASSEMIA TRAIT

S.NO	MRD NO.	IMMUNOPATH NO.	AGE	SEX	PLACE OF BIRTH	CASTE/RELIGION	PALLOR	BLOOD TRANSFUSION RECEIVED	JAUNDICE	LIVER SPAN	SPLEEN SPAN	FAMILY HISTORY	RBC	Hb	MCV	MCH	MCHC	RDW	MENTZER INDEX	MICROCYTES	NORMOCYTES	PENCIL CELLS	TARGET CELLS	HYPOCHROMIA	NORMOCHROMIA	HbF	HbA	HbA2	HbD	Hb S	IMPRESSION
155	606725	4947/2018	25	F	LUDHIANA	HINDU	YES	NO	NO				2.55	8.9	108.5	35.1	32.6	31.3	42.54		PRESENT					0.7	62.8	2.3	34.2		HbD PUNJAB HETEROZYGOUS
156	605271	5062/2018	22	F	BHATINDA	SIKH	NO	YES	NO	INCREASED	INCREASED	ABSENT	3.89	10.1	79.4	26	32.8	29	20.41	PRESENT	PRESENT		PRESENT	PRESENT		23.2	72	4.8			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
157	600620	5078/2018	24	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.65	14.1	70	21.2	30.2	15.6	10.52	PRESENT	PRESENT			PRESENT		1.3	93.3	5.4			BETA THALASSEMIA TRAIT
158	232990	5279/2018	27	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	4.26	8.5	67	20	29.7	15.7	15.72	PRESENT	PRESENT			PRESENT		0.7	94.3	5			BETA THALASSEMIA TRAIT
159	526615	5280/2018	28	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	4.87	11.7	77	24.1	31.4	16.6	15.81	PRESENT	PRESENT			PRESENT		0.7	95.2	4.1			SUGGESTIVE OF BETA THALASSEMIA TRAIT
160	597563	5356/2018	31	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.2	10.1	63	19.5	30.9	15	12.11	PRESENT	PRESENT			PRESENT		0.7	94.1	5.2			BETA THALASSEMIA TRAIT
161	624167	5504/2018	32	F	BARNALA	SIKH	YES	NO	NO				3.09	9.1	88	29.4	33.3	14.2	28.47		PRESENT				PRESENT	0.7	59.3	2.5	37.5		HbD PUNJAB HETEROZYGOUS
162	558198	5521/2018	3	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.18	9.4	57.8	18.2	31.5	18	11.15	PRESENT	PRESENT			PRESENT		1.4	92.8	5.8			BETA THALASSEMIA TRAIT
163	625559	5522/2018	3	M	LUDHIANA	HINDU	NO	NO	NO			PRESENT	5.46	9.5	59	17.4	29.5	15.8	10.8	PRESENT	PRESENT			PRESENT		5.2	89.3	5.5			BETA THALASSEMIA TRAIT
164	630249	5646/2018	25	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.26	12.3	73.9	23.4	31.7	16.7	14.04	PRESENT	PRESENT			PRESENT		0.7	94.1	5.2			BETA THALASSEMIA TRAIT
165	630223	5655/2018	43	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.72	7.3	54	15.4	28.4	20.9	11.44	PRESENT	PRESENT			PRESENT		0	95.9	4.1			SUGGESTIVE OF BETA THALASSEMIA TRAIT
166	629866	5658/2018	3	F	LUDHIANA	SIKH	NO	YES	NO		INCREASED	PRESENT	2.54	4.5	62	17.7	28.5	34.8	24.4	PRESENT	PRESENT	PRESENT				31.7	7.2	61.1			COMPOUND HETEROZYGOUS FOR HbE AND BETA THALASSEMIA
167	631891	5688/2018	32	M	LUDHIANA	SIKH	NO	NO	NO			PRESENT	4.97	12.6	79	25.4	32.2	17.3	15.89	PRESENT	PRESENT			PRESENT		0.7	90.4	8.9			BETA THALASSEMIA TRAIT
168	633369	5733/2018	28	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.31	10.4	62.3	19.5	31.3	21.7	11.73	PRESENT	PRESENT			PRESENT		0.7	94.4	4.9			BETA THALASSEMIA TRAIT
169	612488	5753/2018	25	F	SIRSA	HINDU	NO	NO	NO			ABSENT	5.23	9.8	61	18.7	30.6	16.6	11.66	PRESENT	PRESENT			PRESENT		9.1	85.4	5.5			BETA THALASSEMIA TRAIT
170	834593	5754/2018	27	F	KAITHAL	HINDU	NO	NO	NO			ABSENT	4.4	10	71.6	22.8	31.8	17	16.13	PRESENT	PRESENT			PRESENT		1.4	93.5	5.1			BETA THALASSEMIA TRAIT
171	634634	5755/2018	27	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	4.86	10.3	66.7	21.1	31.6	17.1	13.72	PRESENT	PRESENT			PRESENT		0.7	93.5	5.8			BETA THALASSEMIA TRAIT
172	634759	5756/2018	2	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.53	10.4	59.5	18.7	31.5	17.2	10.75	PRESENT	PRESENT			PRESENT		4.1	90.6	5.3			BETA THALASSEMIA TRAIT
173	635716	5975/2018	17	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	3.24	8.6	87	26.4	30.3	18.5	26.85	PRESENT	PRESENT			PRESENT		1.2	79.4	4.4			BETA THALASSEMIA TRAIT
174	645597	34/2019	31	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.97	10.4	66.6	20.9	31.4	15	13.4	PRESENT	PRESENT			PRESENT		1.6	82.3	5.6			BETA THALASSEMIA TRAIT
175	538093	271/2019	29	F	LUDHIANA	HINDU	NO	NO	NO				4.61	13	87	28.1	32.4	12.9	18.87		PRESENT				PRESENT	0.8	53.4	2.5	36.7		HbD PUNJAB HETEROZYGOUS
176	660822	499/2019	24	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.57	12.1	68.4	21.7	31.7	15.1	12.28	PRESENT	PRESENT				PRESENT	1.4	93	5.6			BETA THALASSEMIA TRAIT
177	6630	570/2019	39	F	LUDHIANA	SIKH	NO	NO	NO				4.26	11.7	85	27.5	32.4	15.1	13.61		PRESENT				PRESENT	0.8	50.3	2.9	36.6		HbD PUNJAB HETEROZYGOUS
178	660777	578/2019	1	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.29	5.9	46.5	13.9	29.8	34.6	10.83	PRESENT	PRESENT	PRESENT			PRESENT	1.8	79.8	4.5			SUGGESTIVE OF BETA THALASSEMIA TRAIT
179	652727	674/2019	50	M	FAZILKA	SIKH	YES	YES	NO			ABSENT	4.75	7.6	57	16.1	28.1	20.6	12	PRESENT	PRESENT		PRESENT	PRESENT		0.8	79.5	4.5			BETA THALASSEMIA TRAIT
180	673349	838/2019	29	F	UNA	HINDU	NO	NO	NO				4.67	12.5	87	26.8	30.9	14.5	18.62		PRESENT				PRESENT	0.8	85.3	3.8			MILDLY RAISED HbA2 LEVELS NOTED
181	673718	893/2019	55	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.28	10.7	64.9	20.2	31.1	15.2	12.29	PRESENT	PRESENT			PRESENT		1.1	79.4	6.2			BETA THALASSEMIA TRAIT
182	675715	899/2019	25	F	LUDHIANA	HINDU	NO	NO	NO				4.13	12.9	80.5	26.3	32.6	15.2	19.49		PRESENT				PRESENT		71.8	2.5			HbQ INDIA HETEROZYGOUS
183	653853	908/2019	30	F	LUDHIANA	SIKH	YES	NO	NO				3.82	10.6	91	27.8	30.5	13.6	23.82		PRESENT				PRESENT	0.8	85.6	3.8			MILDLY RAISED HbA2 LEVELS NOTED
184	336075	939/2019	29	F	NAWANSHAHAR	SIKH	NO	NO	NO				4.55	12.9	85.4	28.3	33.1	12.5	18.76		PRESENT				PRESENT	0.7	85	3.9			BORDERLINE HbA2 LEVELS SEEN
185	589699	1013/2019	27	F	LUDHIANA	SIKH	NO	NO	NO				4.67	11.4	86	24.5	28.6	16.1	18.41		PRESENT				PRESENT	0.7	84.9	3.6			MILDLY RAISED HbA2 LEVELS NOTED
186	680916	1118/2019	42	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.35	11.3	69	21.1	30.4	13.6	12.89	PRESENT	PRESENT			PRESENT		0.8	81.9	6.1			BETA THALASSEMIA TRAIT
187	666499	1147/2019	24	F	LUDHIANA	HINDU	NO	NO	NO				4.54	12.3	84	27.1	32.3	15.1	18.5		PRESENT				PRESENT	0.8	85.4	3.6			MILDLY RAISED HbA2 LEVELS NOTED
188	7931	1152/2019	33	F	FARIDKOT	SIKH	NO	NO	NO				3.94	11.6	86.39	29.3	33.7	14	22.05		PRESENT				PRESENT	0.8	84.6	3.6			MILDLY RAISED HbA2 LEVELS NOTED
189	687268	1269/2019	23	F	BARNALA	SIKH	NO	NO	NO			PRESENT	4.99	11.9	77	23.8	31	14.8	15.43		PRESENT				PRESENT	2.8	79.2	6.4			BETA THALASSEMIA TRAIT
190	688248	1270/2019	32	F	LUDHIANA	HINDU	NO	NO	NO				3.73	12.2	97.4	32.8	33.6	15	26.11		PRESENT				PRESENT	0.8	86.8	3.7			MILDLY RAISED HbA2 LEVELS NOTED
191	687345	1272/2019	33	F	LUDHIANA	SIKH	NO	NO	NO				4.79	13	86	27.1	31.7	15.4	17.95		PRESENT				PRESENT	0.8	85.2	3.6			MILDLY RAISED HbA2 LEVELS NOTED
192	691564	1392/2019	0.166666667	F	PATIALA	HINDU	NO	NO	NO	INCREASED	INCREASED	PRESENT(M	1.71	4.7	80	27.3	34.1	32.8	46.78	PRESENT		PRESENT	PRESENT	PRESENT		95.6	2.3	0.6			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
193		1474/2019	6	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	4.63	11.7	87.9	25.3	28.8	16.5	18.98		PRESENT				PRESENT	2.3	75.6	4.5			SUGGESTIVE OF BETA THALASSEMIA TRAIT
194	697309	1550/2019	44	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.23	5.9	50.6	13.9	27.5	22.7	11.96	PRESENT	PRESENT			PRESENT		0.8	84.2	4.2			BETA THALASSEMIA TRAIT
195	699139	1602/2019	40	M	MANSA	SIKH	NO	YES	YES		INCREASED	ABSENT	1.51	3.3	68.8	21.9	31.7	33.7	45.66							11.3	63.6	10.2			SUGGESTIVE OF BETA THALASSEMIA HETEROZYGOUS
196	454345	1732/2019	32	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.69	12.6	65	18.8	28.9	15.8	9.71	PRESENT	PRESENT			PRESENT		1.2	83.1	6			BETA THALASSEMIA TRAIT
197	389496	1735/2019	34	F	BARNALA	SIKH	NO	NO	NO			ABSENT	4.67	6	50	12.8	25.8	18	10.7	PRESENT				PRESENT		0.7	82	4.5			BETA THALASSEMIA TRAIT
198	701679	1802/2019	60	M	LUDHIANA	JAINSM	NO	NO	NO			ABSENT	7.06	12.4	61	17.5	28.8	16.6	8.64	PRESENT	PRESENT			PRESENT		0.8	79.4	5.5			BETA THALASSEMIA TRAIT
199	61816	1809/2019	26	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.72	8.4	59	14.7	25	18.4	10.31	PRESENT	PRESENT			PRESENT		0.8	82.6	6.4			BETA THALASSEMIA TRAIT
200	704185	1810/2019	25	F	HANUMANGARH	SIKH	YES	NO	NO			ABSENT	4.27	9.6	74	22.4	30.4	19.9	17.33	PRESENT	PRESENT			PRESENT		1.2	80.7	5.9			BETA THALASSEMIA TRAIT
201	705886	1933/2019	26	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.07	12	62.8	19.8	31.5	17.2	11.33	PRESENT	PRESENT			PRESENT		0.8	83.6	5.4			BETA THALASSEMIA TRAIT
202	577416	1938/2019	24	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.5	12.9	74.4	23.4	31.4	15.5	13.52		PRESENT				PRESENT	0.8	84.3	6.1			BETA THALASSEMIA TRAIT
203	706880	1984/2019	67	F	MOGA	HINDU	YES	NO	NO			ABSENT	5.47	9.2	56.3	16.8	29.8	19.4	10.29	PRESENT	PRESENT			PRESENT		0	77.2	6.1			BETA THALASSEMIA TRAIT
204	705205	1985/2019	61	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.33	10.2	67	19.2	28.8	17.4	12.57	PRESENT	PRESENT			PRESENT		3.7	77.9	5.4			BETA THALASSEMIA TRAIT
205	708573	1994/2019	30	F	KAPURTHALA	HINDU	NO	NO	NO			ABSENT	4.11	8.6	66.6	20.8	31.2	15.5	16.2	PRESENT	PRESENT			PRESENT		0.8	83.2	5.7			BETA THALASSEMIA TRAIT
206	210147	2053/2019	27	F	JALANDHAR	SIKH	YES	NO	NO			ABSENT	4.94	9.9	64.3	20.1	31.3	22.6	13.01	PRESENT	PRESENT			PRESENT		1.2	82.6	5.6			BETA THALASSEMIA TRAIT
207	8413	2058/2019	28	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	7.12	12.9	65	18.1	27.7	17.4	9.12	PRESENT	PRESENT			PRESENT		0.8	81.1	6.7			BETA THALASSEMIA TRAIT
208	370624	2170/2019	35	M	MOGA	SIKH	NO	NO	NO			ABSENT	6.14	11.4	60.6	18.6	30.8	16.6	9.86	PRESENT	PRESENT			PRESENT		0.8	81.6	6.6			BETA THALASSEMIA TRAIT
209	604599	2253/2019	47	M	UDHAMPUR	HINDU	NO	NO	NO			ABSENT	5.81	11.3	60.9	19.5	31.9	17.3	10.48	PRESENT	PRESENT			PRESENT		0.8	83.2	5.1			BETA THALASSEMIA TRAIT
210	719270	2379/2019	1	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.46	9.5	55.6	17.5	31.5	22.8	10.18	PRESENT	PRESENT		PRESENT	PRESENT		6.2	77.1	5.7			SUGGESTIVE OF BETA THALASSEMIA TRAIT
211	720081	2386/2019	21	F	LUDHIANA	SIKH	YES	YES	YES			PRESENT	1.41	2.7	66	19.1	29	31	46.8	PRESENT	PRESENT	PRESENT		PRESENT		52.5	31.3	8.2			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
212	1110	2487/2019	17	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.79	13.6	63.3	20	31.7	15.8	9.32	PRESENT	PRESENT			PRESENT		1.3	81.2	5.8			BETA THALASSEMIA TRAIT
213	723633	2545/2019	60	F	JALANDHAR	HINDU	YES	YES	YES			ABSENT	2	5.8	91.2	29.2	32.1	24.9	45.6	PRESENT	PRESENT	PRESENT	PRESENT	PRESENT		20.1	64.1	4.1			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
214	597389	2672/2019	18	M	LUDHIANA	SIKH	YES	NO	NO				2.44	8.9	112	36.7	32.8	16.4	45.9		PRESENT					0.8	55.2	3.3	35.9		HbD PUNJAB HETEROZYGOUS
215	729418	2736/2019	26	F	LUDHIANA	HINDU	YES	NO	NO				3.26	8.1	80.7	24.8	30.7	16.5	24.75	PRESENT	PRESENT			PRESENT		0.8	83.5	4			MILDLY RAISED HbA2 LEVELS NOTED
216	730103	2740/2019	27	M	LUDHIANA	SIKH	NO	NO	NO				5.98	16.2	82.8	27	32.7	15.8	13.84		PRESENT				PRESENT	0.8	53.3	2.7	35		HbD PUNJAB HETEROZYGOUS
217	563384	2815/2019	29	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.24	10	61.7	19	30.8	15.9	11.77							0.8	83.4	5.4			BETA THALASSEMIA TRAIT
218	731499	2825/2019	27	F	LUDHIANA	SIKH	YES	YES	NO				1.75	3.9	70.4	22.2	31.5	22.2	40.22	PRESENT	PRESENT		PRESENT	PRESENT		0.8	56.7	2.7	32.9		HbD PUNJAB HETEROZYGOUS
219	730810	2826/2019	31	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.93	7	51.1	14.1	27.7	23.1	10.36	PRESENT	PRESENT			PRESENT		1	81.7	5.6			BETA THALASSEMIA TRAIT
220	732125	2827/2019	8	M	LUDHIANA	SIKH	YES	NO	NO	INCREASED	INCREASED	PRESENT	3.9	7.8	66.3	19.9	30	30.3	17	PRESENT	PRESENT		PRESENT	PRESENT		87.1	7.9	3.7			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
221	728879	2893/2019	26	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.39	9.2	66.2	20.9	31.6	22.5	15.07	PRESENT	PRESENT		PRESENT	PRESENT		0.8	81.9	6.1			BETA THALASSEMIA TRAIT
222	736010	2982/2019	28	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5	9.6	63.7	19.1	30	18.4	12.74	PRESENT	PRESENT			PRESENT		1	82.8	6.4			BETA THALASSEMIA TRAIT
223	732303	2983/2019	9	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	6.09	10.3	59	16.9	28.5	16.8	9.68	PRESENT	PRESENT			PRESENT		0.8	82.9	5.3			BETA THALASSEMIA TRAIT
224	52807	2984/2019	28	F	LUDHIANA	SIKH	YES	NO	NO			PRESENT	5.72	10.6	59.2	18.6	31.4	16.5	10.34	PRESENT	PRESENT			PRESENT		0	82.2	5.3			BETA THALASSEMIA TRAIT
225	735208	2986/2019	32	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.96	11.8	63.2	19.8	31.3	17	10.6	PRESENT	PRESENT			PRESENT		2.9	80.6	5.1			BETA THALASSEMIA TRAIT
226	735464	2990/2019	29	M	LUDHIANA	SIKH	NO	NO	NO			PRESENT	6.7	13.3	67	19.8	29.4	18.3	10	PRESENT	PRESENT			PRESENT		0.8	80.5	6.3			BETA THALASSEMIA TRAIT
227	701517	2993/2019	0.333333333	M	LUDHIANA	SIKH	YES	YES	NO	INCREASED	INCREASED	PRESENT(25	2.56	5.3	62	20.7	33.1	26.3	24.21	PRESENT	PRESENT	PRESENT		PRESENT		92	2.2	2.1			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
228	699811	2994/2019	60	M	LUDHIANA	HINDU	YES	YES	YES			ABSENT	4.3	9.3	69.7	21.6	30.9	18.2	16.2	PRESENT	PRESENT			PRESENT		4.5	73.3	4.6			BETA THALASSEMIA TRAIT
229	741485	3139/2019	37	F	JALANDHAR	HINDU	NO	NO	NO				4.48	13.1	87.3	29.2	33.4	15.7	19.48		PRESENT				PRESENT	0.8	51	2.5	37.9		HbD PUNJAB HETEROZYGOUS
230	151027	3223/2019	52	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	4.82	9.4	69	19.4	28.1	16.2	14.31	PRESENT	PRESENT			PRESENT		1.9	78	7.7			BETA THALASSEMIA TRAIT
231	744229	3296/2019	32	F	BARNAKA	SIKH	NO	NO	NO			ABSENT	4.67	10.9	73.4	23.3	31.7	17.8	15.71	PRESENT	PRESENT			PRESENT		1.3	80.4	6.5			BETA THALASSEMIA TRAIT

S.NO	MRD NO.	IMMUNOPATH NO.	AGE	SEX	PLACE OF BIRTH	CASTE/RELIGION	PALLOR	BLOOD TRANSFUSION RECEIVED	JAUNDICE	LIVER SPAN	SPLEEN SPAN	FAMILY HISTORY	RBC	Hb	MCV	MCH	MCHC	RDW	MENTZER INDEX	MICROCYTES	NORMOCYTES	PENCIL CELLS	TARGET CELLS	HYPOCHROMIA	NORMOCHROMIA	HbF	HbA	HbA2	HbD	Hb S	IMPRESSION
232	742669	3307/2019	18	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	3.82	7.5	79.9	24.8	31	16.4	26.45	PRESENT	PRESENT			PRESENT		0.8	86	4.3			BETA THALASSEMIA TRAIT
233	734276	3355/2019	19	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.46	11.8	63	18.3	29.1	16.8	9.75		PRESENT			PRESENT		0.8	81.2	6.8			BETA THALASSEMIA TRAIT
234	749690	3487/2019	26	M	LUDHIANA	HINDU	YES	NO	YES	INCREASED			5.49	12	71.1	21.9	38.8	22.4	12.93					PRESENT		3.3	81.7	5.5			COMPOUND HETEROZYGOUS OF HbQ AND BETA THALASSEMIA
235	514702	3492/2019	25	F	LUDHIANA	SIKH	NO	NO	NO				4.13	12.7	90.7	30.6	33.8	14.4	21.96		PRESENT				PRESENT	0.8	56.8	2.7	36.6		HbD PUNJAB HETEROZYGOUS
236	374825	3494/2019	49	M	LUDHIANA	SIKH	YES	YES	YES		INCREASED		2.81	6.8	78	24.1	30.9	26.3	27.7	PRESENT	PRESENT			PRESENT		0.8	82	4.8			SUGGESTIVE OF HETEROZYGOUS BETA THALASSEMIA
237	618016	3495/2019	26	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.1	8.6	56	16.8	29.9	19.4	10.98	PRESENT	PRESENT			PRESENT		0.8	82.3	6			BETA THALASSEMIA TRAIT
238	729758	3497/2019	28	F	LUDHIANA	SIKH	YES	NO	NO				4.95	9.5	66	19.3	29.3	24.4	13.33	PRESENT	PRESENT			PRESENT			62.1	40.9			HbD IRAN HETEROZYGOUS
239	750691	3528/2019	32	F	KAPURTHALA	SIKH	YES	NO	NO			PRESENT	5.22	9.4	60	18	30	18.5	11.49	PRESENT	PRESENT			PRESENT		1.1	80.6	4.7			BETA THALASSEMIA TRAIT
240	754171	3603/2019	10	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.98	10.1	58	16.8	29	16.4	9.69	PRESENT	PRESENT			PRESENT		2	79.7	5.9			BETA THALASSEMIA TRAIT
241	754391	3607/2019	10	F	LUDHIANA	HINDU	YES	NO	NO				6.77	10.8	55	16	28	20	8.12	PRESENT	PRESENT		PRESENT	PRESENT		5.8	76.4	6			SUGGESTIVE OF HETEROZYGOUS BETA THALASSEMIA
242	756657	3688/2019	16	F	JAMMU	HINDU	NO	NO	NO				2.87	8.7	93	30.1	32.3	16.8	32.4		PRESENT				PRESENT	24.5	5.3	1.6		66	SUGGESTIVE OF HbS HOMOZYGOUS
243	759401	3811/2019	27	F	JAMMU	HINDU	YES	NO	NO			ABSENT	4.13	9.2	76	22.4	29.3	18.1	18.4	PRESENT	PRESENT			PRESENT		0.8	83.6	4.6			BETA THALASSEMIA TRAIT
244	559727	3812/2019	31	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.49	9.9	62	18.1	29	17.3	11.29	PRESENT	PRESENT			PRESENT		0.8	81.7	5			BETA THALASSEMIA TRAIT
245	761703	3854/2019	3	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.42	10.4	64	19.2	30	17.4	11.8	PRESENT	PRESENT			PRESENT		6.2	76.4	5.3			BETA THALASSEMIA TRAIT
246	761025	3860/2019	22	F	LUDHIANA	HINDU	YES	YES	YES				2.35	5	72	21.4	29.8	29	30.63	PRESENT	PRESENT	PRESENT		PRESENT		0.8	75.6	2.8	11.7		HbD PUNJAB HETEROZYGOUS
247	765393	3939/2019	28	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.56	9.4	59	16.9	28.4	16.3	10.61	PRESENT	PRESENT			PRESENT		0.8	80.6	5.7			BETA THALASSEMIA TRAIT
248	490641	3940/2019	26	F	HOSHIARPUR	HINDU	YES	NO	NO			ABSENT	3.98	9.5	78	23.8	30.7	18	19.59	PRESENT	PRESENT			PRESENT		4.1	81.2	4.9			BETA THALASSEMIA TRAIT
249	530289	3945/2019	27	F	LUDHIANA	SIKH	NO	NO	NO				4.18	12.2	90	29.2	32.3	15.4	21.53		PRESENT				PRESENT		86.1	3.6			MILDLY RAISED HbA2 LEVELS NOTED
250	744676	4007/2019	55	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	6.15	11	62	17.9	28.9	15.6	10.08	PRESENT	PRESENT			PRESENT		0.8	82.2	5.9			BETA THALASSEMIA TRAIT
251	769062	4072/2019	26	F	LUDHIANA	HINDU	NO	NO	NO				4.07	11.3	87	27.8	32.1	15.5	2.13		PRESENT				PRESENT	0.8	53.6	44.5			HbD IRAN HETEROZYGOUS
252	768305	4081/2019	26	F	LUDHIANA	HINDU	YES	NO	NO		INCREASED	ABSENT	4.52	9.1	68	20.2	29.8	17.7	15.04	PRESENT	PRESENT			PRESENT		0.8	58.6	3.2		29.9	HbS HETEROZYGOUS
253	771654	4139/2019	30	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.64	10.9	60.7	19.2	31.7	16.4	10.76	PRESENT	PRESENT			PRESENT		0.9	82	5.6			BETA THALASSEMIA TRAIT
254	6885	4261/2019	35	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.78	10.7	69.6	22.3	32.1	18	14.56	PRESENT	PRESENT			PRESENT		0.9	81.4	6.3			BETA THALASSEMIA TRAIT
255	778641	4373/2019	39	M	LUDHIANA	HINDU	NO	NO	NO			PRESENT	6.73	13.2	66	19.6	29.5	16.2	9.8	PRESENT	PRESENT			PRESENT		1.2	80.2	5			BETA THALASSEMIA TRAIT
256	765124	4378/2019	10	F	LUDHIANA	SIKH	NO	NO	NO			PRESENT	5.44	11.4	68	25	30.9	16.6	12.5	PRESENT	PRESENT			PRESENT		0.8	80.5	6.3			BETA THALASSEMIA TRAIT
257	778642	4380/2019	28	F	LUDHIANA	HINDU	YES	YES	YES			PRESENT	4.92	9.5	64	19.3	30	17.3	13	PRESENT	PRESENT			PRESENT		3.9	7	4.5	78.5		COMPOUND HETEROZYGOUS OF HbD PUNJABAND BETA THALASSEMIA
258	783014	4515/2019	59	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.78	8.3	58	17.3	30	15.4	12.13	PRESENT	PRESENT			PRESENT		0	78.1	5.4			BETA THALASSEMIA TRAIT
259	785155	4519/2019	25	F	LUDHIANA	SIKH	YES	YES	YES			ABSENT	2.7	5.6	71.2	20.8	29.3	23.8	26.37	PRESENT	PRESENT			PRESENT		1.6	79.5	5.6			BETA THALASSEMIA TRAIT
260	782700	4521/2019	29	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.92	11.3	64	19.1	30	15.9	10.81	PRESENT	PRESENT			PRESENT		0.8	82.7	5.2			BETA THALASSEMIA TRAIT
261	783060	4522/2019	35	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.15	13.3	68.6	21.6	31.6	17.3	11.15	PRESENT	PRESENT			PRESENT		2.1	81.7	5.1			BETA THALASSEMIA TRAIT
262	646476	4606/2019	29	F	LUDHIANA	SIKH	NO	NO	NO				4.09	12.7	93.2	30.9	33.2	13.4	22.78		PRESENT				PRESENT		83.4	3.8			MILDLY RAISED HbA2 LEVELS NOTED
263	323906	4675/2019	17	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.08	11.8	62.3	19.4	31.2	18.1	10.24	PRESENT	PRESENT			PRESENT		0.8	81.7	6.9			BETA THALASSEMIA TRAIT
264	791064	4764/2019	26	F	BARNALA	SIKH	NO	NO	NO				4.28	12.7	89	29.7	33.3	13.3	20.79		PRESENT				PRESENT	0	56.4	2.4	36.2		HbD PUNJAB HETEROZYGOUS
265	792917	4837/2019	22	M	FATEHGARH SAHIB	HINDU	YES	YES	YES		INCREASED		0.93	2.4	87.1	26	29.9	24.7	93.54	PRESENT	PRESENT		PRESENT	PRESENT		35.4	18.3	4		34.9	COMPOUND HETEROZYGOUS OF HbS AND BETA THALASSEMIA
266	790152	4838/2019	59	F	LUDHIANA	HINDU	YES	YES	NO			ABSENT	3.62	6.3	56	17.3	31.2	16.3	15.46	PRESENT	PRESENT	PRESENT		PRESENT		0.78	77	5.4			BETA THALASSEMIA TRAIT
267	799773	5022/2019	29	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.8	12.3	78	25.6	32.7	14.7	16.25	PRESENT	PRESENT			PRESENT		0.8	84.6	4			SUGGESTIVE OF BETA THALASSEMIA TRAIT
268	800400	5027/2019	30	F	LUDHIANA	HINDU	YES	NO	NO			PRESENT	5.07	13	81	25.5	31.5	15.4	15.97		PRESENT				PRESENT	0	84.6	4			SUGGESTIVE OF BETA THALASSEMIA TRAIT
269	799379	5029/2019	49	F	PATHANKOT	SIKH	YES	YES	NO			ABSENT	2.93	6.6	72	22.4	31.3	16.6	24.57	PRESENT	PRESENT			PRESENT		1.2	81.3	5.6			SUGGESTIVE OF BETA THALASSEMIA TRAIT
270	404036	5034/2019	26	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5	10.1	67	20.3	30.2	16.2	13.4	PRESENT	PRESENT			PRESENT		1	82	5.5			SUGGESTIVE OF BETA THALASSEMIA TRAIT
271	802911	5113/2019	30	F	LUDHIANA	SIKH	NO	NO	NO				4	12.7	93.4	31.8	34	12.8	23.35		PRESENT				PRESENT	0.8	85.8	3.7			BORDERLINE HbA2 LEVELS SEEN
272	780034	5121/2019	30	F	LUDHIANA	HINDU	NO	NO	NO				4.36	12.8	91	29.5	32.5	15.2	20.87		PRESENT				PRESENT	0.8	72.2	2.8			HbQ INDIA HETEROZYGOUS
273	802962	5123/2019	18	F	JALANDHAR	SIKH	YES	NO	NO				2.82	8	95	31	32	18.1	33.68		PRESENT						85.1	3.7			BORDERLINE HbA2 LEVELS SEEN
274	401871	5161/2019	32	F	LUDHIANA	HINDU	YES	NO	NO				4.69	10.3	69.8	21.9	31.4	16.6	14.88	PRESENT	PRESENT			PRESENT			83.5	3.7			BORDERLINE HbA2 LEVELS SEEN
275	805845	5172/2019	35	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	6.64	12.2	61	18.4	30.2	16		PRESENT	PRESENT			PRESENT		1.6	80.7	7.1			BETA THALASSEMIA TRAIT
276	804845	5176/2019	29	F	LUDHIANA	HINDU	NO	NO	NO			PRESENT	5.25	14.9	83.2	28.4	34.1	13.7	15.84		PRESENT			PRESENT		0	82.3	4.5			BETA THALASSEMIA TRAIT
277	792523	5179/2019	24	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.01	8.5	72	21.1	29.2	18.8	17.95	PRESENT	PRESENT			PRESENT		1.2	82.5	6.7			BETA THALASSEMIA TRAIT
278	810267	5308/2019	2	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.44	6.4	46	11.8	25.8	20.4	8.45	PRESENT	PRESENT	PRESENT		PRESENT		0.8	81.7	4.5			BETA THALASSEMIA TRAIT
279	812920	5356/2019	25	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	3.47	7.3	68	21.1	31	16.3	19.59	PRESENT	PRESENT					0.8	82.9	5.5			BETA THALASSEMIA TRAIT
280	813767	5460/2019	26	F	FEROZPUR	SIKH	NO	NO	NO				4.22	12.7	88	30.2	34.3	15.7	20.85		PRESENT				PRESENT	0.8	83	3.2	36.5		HbD PUNJAB HETEROZYGOUS
281	815910	5469/2019	40	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.35	10.8	68	20.2	29.8	17	12.71	PRESENT	PRESENT			PRESENT		1.6	81.3	5.7			BETA THALASSEMIA TRAIT
282	808266	5470/2019	48	F	LUDHIANA	JAINSM	YES	NO	NO			ABSENT	5.46	10.2	61.8	18.7	30.3	16.7	11.31	PRESENT	PRESENT			PRESENT		0.9	80.6	5.2			BETA THALASSEMIA TRAIT
283	819135	5586/2019	21	F	LUDHIANA	SIKH	NO	NO	NO				4.62	13.5	88	29.2	33	14.2	19.04		PRESENT				PRESENT	0.8	49.8	3.1	37		HbD PUNJAB HETEROZYGOUS
284	818885	5638/2019	32	M	JAMMU	HINDU	NO	YES	YES				1.77	6.8	119.3	38.2	32	15.2	67.4		PRESENT						82.6	3.7			BORDERLINE HbA2 LEVELS SEEN
285	820399	5639/2019	28	F	AMRITSAR	SIKH	YES	YES	NO				2.39	6.2	82.5	25.9	31.4	17.1	34.51	PRESENT	PRESENT			PRESENT		0.8	85.6	3.7			BORDERLINE HbA2 LEVELS SEEN
286	821964	5714/2019	21	F	LUDHIANA	HINDU	NO	NO	NO				3.69	12.2	99.2	33	33.3	16.6	26.88		PRESENT				PRESENT	0.8	86.1	3.6			MILDLY RAISED HbA2 LEVELS NOTED
287	802546	5717/2019	30	M	LUDHIANA	HINDU	NO	NO	NO				5.2	16.3	95	31.3	33.1	15.5	18.26		PRESENT				PRESENT		85.2	3.8			MILDLY RAISED HbA2 LEVELS NOTED
288	823187	5720/2019	21	F	LUDHIANA	HINDU	NO	NO	NO				4.09	12.5	93	30.5	32.8	13.2	22.73		PRESENT				PRESENT	0.8	55.2	3.4	37.5		HbD PUNJAB HETEROZYGOUS
289	824899	5777/2019	29	F	LUDHIANA	HINDU	YES	NO	NO				3.8	11.2	88	29	33.2	15.7	22.91		PRESENT				PRESENT	0.8	85.2	3.6			BORDERLINE HbA2 LEVELS SEEN
290	826476	5845/2019	28	M	LUDHIANA	SIKH	NO	NO	NO			PRESENT	6.03	12.3	64.5	20.4	31.6	17.2	10.69	PRESENT	PRESENT			PRESENT		1.1	81.1	5.2			BETA THALASSEMIA TRAIT
291	826326	5846/2019	26	F	LUDHIANA	SIKH	NO	NO	NO			PRESENT	5.22	11.3	69.5	21.6	31.1	16.4	13.31	PRESENT	PRESENT			PRESENT		4.2	78.8	5.5			BETA THALASSEMIA TRAIT
292	810971	5850/2019	18	F	LUDHIANA	HINDU	YES	NO	NO			PRESENT	4.67	9.3	67	20	30	18.7	14.34	PRESENT	PRESENT			PRESENT		1.1	81.2	6.1			BETA THALASSEMIA TRAIT
293	828791	5890/2019	8	M	SANGRUR	HINDU	YES	NO	NO			PRESENT	6.3	10.3	56.2	17	30.3	17.2	9.32	PRESENT	PRESENT			PRESENT		0.8	80.4	5.4			BETA THALASSEMIA TRAIT
294	829793	5891/2019	10	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.73	10.4	59.2	18.1	30.6	16.9	11.02	PRESENT	PRESENT			PRESENT		0.8	81.2	5.3			BETA THALASSEMIA TRAIT
295	786835	5960/2019	22	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.74	12.2	68.9	21.2	30.8	16.2	12	PRESENT	PRESENT			PRESENT		2.6	79.8	6.1			BETA THALASSEMIA TRAIT
296	831738	5961/2019	26	F	BARNALA	HINDU	YES	NO	NO			ABSENT	3.91	8.6	70	22	31.5	17.1	17.9	PRESENT	PRESENT			PRESENT		1	81.3	5.5			BETA THALASSEMIA TRAIT
297	832286	6009/2019	33	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.36	10.4	65	19.5	30.1	16.9	12.12	PRESENT	PRESENT			PRESENT		0.8	82	5.6			BETA THALASSEMIA TRAIT
298	832944	6010/2019	31	F	LUDHIANA	HINDU	YES	NO	NO			PRESENT	4.78	10	67.5	21	31	16.5	8.67	PRESENT	PRESENT			PRESENT		1.9	81.6	5.4			BETA THALASSEMIA TRAIT
299	329217	6016/2019	72	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	3.17	7	72	21.9	30.5	20.9	22.71	PRESENT	PRESENT			PRESENT		1.2	82.3	5.1			BETA THALASSEMIA TRAIT
300	832454	6018/2019	27	M	JAMMU	SIKH	YES	NO	NO			ABSENT	2.32	9.3	119	39.9	33.6	22.3	51.29		PRESENT		PRESENT			2.1	82.6	4			BETA THALASSEMIA TRAIT
301	835854	6104/2019	38	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.07	11.9	66	19.7	29.9	16.2	10.87	PRESENT	PRESENT			PRESENT		1.3	80.9	5.1			BETA THALASSEMIA TRAIT
302	823208	6243/2019	28	F	LUDHIANA	HINDU	NO	NO	NO				3.8	11.3	90	29.7	33.1	15.2	23.68		PRESENT				PRESENT	1	84.8	3.8			BORDERLINE HbA2 LEVELS SEEN
303	840537	6244/2019	36	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	6.82	12.2	61	17.8	29.2	17.7	8.94	PRESENT	PRESENT			PRESENT		2.4	80.2	5.2			BETA THALASSEMIA TRAIT
304	7877	6254/2019	35	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	3.66	13.8	77	24.4	31.7	16.6	13.6	PRESENT	PRESENT			PRESENT		3.2	75.3	6.3			BETA THALASSEMIA TRAIT
305	830993	6411/2019	54	F	LUDHIANA	HINDU	YES	NO	NO				4.32	9.2	70	21.3	30.5	18	16.2	PRESENT	PRESENT			PRESENT		0.8	57.8	2.3	31.9		HbD PUNJAB HETEROZYGOUS
306	356921	6526/2019	28	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.26	10.3	76	24.1	31.5	16.1	17.84	PRESENT	PRESENT			PRESENT		8.7	74.3	5.4			BETA THALASSEMIA TRAIT
307	849923	6527/2019	27	F	LUDHIANA	SIKH	NO	NO	NO				3.77	11	90	29.2	32.3	13.4	23.87	PRESENT	PRESENT					0.8	86.1	3.8			BORDERLINE HbA2 LEVELS SEEN
308	39723	6528/2019	31	F	MOGA	SIKH	NO	NO	NO				4.37	12.1	85	27.6	32.6	15.2	19.45		PRESENT				PRESENT	0	53.1	2.6	36.4		HbD PUNJAB HETEROZYGOUS

S.NO	MRD NO.	IMMUNOPATH NO.	AGE	SEX	PLACE OF BIRTH	CASTE/RELIGION	PALLOR	BLOOD TRANSFUSION RECEIVED	JAUNDICE	LIVER SPAN	SPLEEN SPAN	FAMILY HISTORY	RBC	Hb	MCV	MCH	MCHC	RDW	MENTZER INDEX	MICROCYTES	NORMOCYTES	PENCIL CELLS	TARGET CELLS	HYPOCHROMIA	NORMOCHROMIA	HbF	HbA	HbA2	HbD	Hb S	IMPRESSION
309	850796	6536/2019	33	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.38	11.9	59.8	18.6	31.1	16.4	9.37	PRESENT	PRESENT			PRESENT		0.8	82.4	5.1			BETA THALASSEMIA TRAIT
310	853788	6617/2019	29	F	BARNALA	SIKH	NO	NO	NO			ABSENT	5.09	12.2	78	24	30.9	14.4	15.32		PRESENT				PRESENT	2.2	80.3	5.6			BETA THALASSEMIA TRAIT
311	854069	6631/2019	31	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.95	12.7	62	18.3	29.7	16.3	3.92	PRESENT	PRESENT			PRESENT		0.8	81.3	4.8			BETA THALASSEMIA TRAIT
312	853631	6632/2019	0.583333333	M	BARNALA	SIKH	YES	YES	NO	INCREASED	INCREASED	PRESENT	2.85	5.9	68	20.5	30.1	26.7	23.85	PRESENT	PRESENT	PRESENT	PRESENT	PRESENT		80.9	13.1	2.3			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
313	854430	6635/2019	45	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	7.06	12.8	61	18.2	29.6	17.5	8.64	PRESENT	PRESENT			PRESENT		0.8	79.2	6.6			BETA THALASSEMIA TRAIT
314	857182	6754/2019	57	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	6.72	13.1	64.2	19.5	30.4	15.7	9.55	PRESENT	PRESENT			PRESENT		0	83.6	5.6			BETA THALASSEMIA TRAIT
315	862721	6911/2019	32	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	3.44	11.3	69	20.8	30.2	17	12.68	PRESENT	PRESENT			PRESENT		1	82.6	5.1			BETA THALASSEMIA TRAIT
316	825432	6912/2019	23	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.52	13.6	69	20.9	30.2	15.4	10.58	PRESENT	PRESENT			PRESENT		2.5	80.1	6.4			BETA THALASSEMIA TRAIT
317	641577	7037/2019	18	M	LUDHIANA	SIKH	NO	NO	NO				6.07	16.3	79.8	27.2	34.1	13.2	13.14		PRESENT				PRESENT	0.8	53.9	2.6	35.3		HbD PUNJAB HETEROZYGOUS
318	872712	7269/2019	71	M	LUDHIANA	SIKH	YES	NO	YES			ABSENT	3.62	7.4	66.1	20.6	31.2	17.4	18.25	PRESENT	PRESENT			PRESENT		0.8	81.9	5.1			BETA THALASSEMIA TRAIT
319	854677	7271/2019	32	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.58	9.9	70	21.7	31	16.4	15.28	PRESENT	PRESENT			PRESENT		0.8	80.6	5			BETA THALASSEMIA TRAIT
320	146524	7277/2019	37	F	LUDHIANA	HINDU	NO	NO	NO				5.35	12.8	75	23.9	31.8	15.1	40		PRESENT				PRESENT	0.8	58.1	2.4	33.1		HbD PUNJAB HETEROZYGOUS
321	850718	2/2020.	31	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	4.64	9.8	69	21.2	30.9	16.5	14.87	PRESENT				PRESENT		0.8	82.3	5			BETA THALASSEMIA TRAIT
322	874858	3/2020.	30	F	MANSA	SIKH	YES	NO	NO			ABSENT	4.15	9.8	74.7	23.7	31.7	20.8	18.5	PRESENT				PRESENT		3.6	79.5	6.1			BETA THALASSEMIA TRAIT
323	440528	39/2020.	28	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.14	10	64	19.5	30.6	16.4	12.45	PRESENT	PRESENT			PRESENT		3.9	79.8	4.9			BETA THALASSEMIA TRAIT
324	8224	40/2020.	30	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.64	8.2	60	17.6	29.3	16.2	12.93	PRESENT	PRESENT			PRESENT		0.8	82.9	5.1			BETA THALASSEMIA TRAIT
325	846724	50/2020	0.666666667	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.38	9	57	16.7	29.6	17.5	10.59	PRESENT	PRESENT		PRESENT	PRESENT		11.7	71.4	4.2			BETA THALASSEMIA TRAIT
326	880444	66/2020	33	M	LUDHIANA	HINDU	NO	NO	NO			ABSENT	6.44	13.1	65.1	20.3	31.2	15.7	10	PRESENT	PRESENT			PRESENT		0.8	82.4	5.9			BETA THALASSEMIA TRAIT
327	836301	75/2020	23	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.6	9.6	67	20.9	31.1	16.7	14.56		PRESENT		PRESENT	PRESENT		2.3	82.7	4.4			BETA THALASSEMIA TRAIT
328	395910	125/2020	67	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	4.86	10	64.7	20.5	31.7	26.9	13.31							0	81.3	4.5			BETA THALASSEMIA TRAIT
329	891457	127/2020	14	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.92	8.6	52	14.5	28.1	18.9	8.78							0.8	80.6	4.3			BETA THALASSEMIA TRAIT
330	894602	142/2020	35	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.56	8.5	62	18.6	29.9	16.7	13.59	PRESENT	PRESENT			PRESENT		1.2	81.8	4.5			BETA THALASSEMIA TRAIT
331	214399	143/2020	56	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.49	8.6	62.9	19.2	30.6	15.8	14	PRESENT	PRESENT			PRESENT		1	76.6	4.8			BETA THALASSEMIA TRAIT
332	728052	161/2020	31	F	LUDHIANA	HINDU	YES	NO	NO				3.93	7.9	65.6	20	30.5	15.8	16.69	PRESENT	PRESENT			PRESENT		0.9	83.3	4			BORDERLINE HbA2 LEVELS
333	358211	167/2020	23	F	LUDHIANA	HINDU	NO	NO	NO				4.25	14.2	91	30	33	14.5	19.15		PRESENT				PRESENT	0.8	85.1	3.9			BORDERLINE HbA2 LEVELS
334	898876	174/2020	28	M	LUDHIANA	HINDU	YES	YES	YES				1.17	3.3	91	28.3	31	31.2	77.77	PRESENT	PRESENT		PRESENT	PRESENT		29.1	2.2	3.2		50.6	SUGGESTIVE OF COMPOUND HETEROZYGOUS FOR HbS AND DELTA BETA THALASSEMIA OR HPFH
335	35126	183/2020	15	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.23	10.9	66.6	20.8	31.3	17.5	12.73	PRESENT	PRESENT			PRESENT		2.5	79	6.1			BETA THALASSEMIA TRAIT
336	904062	201/2020	37	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.62	11.3	65.4	20.2	30.8	16.8	11.63	PRESENT	PRESENT			PRESENT		1.8	81.7	5.4			BETA THALASSEMIA TRAIT
337	608853	206/2020	64	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	5.56	10.9	66	19.6	29.9	16	11.87	PRESENT	PRESENT			PRESENT		1.7	79	5.5			BETA THALASSEMIA TRAIT
338	903596	207/2020	53	M	FEROZPUR	HINDU	NO	NO	NO				3.98	12.2	93.5	30.6	32.7	12	23.49		PRESENT				PRESENT		67.9	3.7			HbQ INDIA HETEROZYGOUS WITH QUESTIONABLE BETA THALASSEMIA TRAIT
339	637126	220/2020	57	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	6.24	9.9	60.8	18.8	30.9	19.1	9.74	PRESENT	PRESENT			PRESENT		0	74.7	4			BETA THALASSEMIA TRAIT
340	908511	221/2020	27	F	BATHINDA	SIKH	NO	NO	NO				4.95	13.9	82.9	28.1	33.7	18.7	16.74		PRESENT				PRESENT	0	52.7	2.6	36.4		HbD PUNJAB HETEROZYGOUS
341	906807	229/2020	34	M	JAMMU	MUSLIM	YES	NO	NO			ABSENT	4.52	8.3	60	18.3	30.6	17.5	13.27	PRESENT	PRESENT			PRESENT		1.3	80.3	5.2			BETA THALASSEMIA TRAIT
342	902394	234/2020	35	M	LUDHIANA	SIKH	YES	NO	NO			ABSENT	6.05	10.7	60	17.7	29.5	15.8	9.91	PRESENT	PRESENT			PRESENT		1	80.6	5.9			BETA THALASSEMIA TRAIT
343	910952	235/2020	28	M	LUDHIANA	JAINSM	YES	NO	NO			PRESENT	5.5	11.5	70.1	20.9	29.9	17	12.74	PRESENT	PRESENT			PRESENT		0.8	82	6			BETA THALASSEMIA TRAIT
344	910953	236/2020	28	F	LUDHIANA	JAINSM	YES	NO	NO			PRESENT	3.9	5.3	63.9	23.9	19.9	35	16.5	PRESENT	PRESENT			PRESENT		0.8	82.8	5.6			BETA THALASSEMIA TRAIT
345	890540	263/2020	33	F	SANGRUR	MUSLIM	YES	NO	NO				4.93	13.5	86	27.4	32	14.9	17.44		PRESENT				PRESENT	0.8	52.2	2.7	37.1		HbD PUNJAB HETEROZYGOUS
346	916923	281/2020	38	M	LUDHIANA	SIKH	YES	NO	YES			ABSENT	5.83	10.6	62	18.2	29.5	16.1	10.63	PRESENT	PRESENT			PRESENT		1.5	80.4	5.5			BETA THALASSEMIA TRAIT
347	151845	284/2020	33	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.74	10.3	62	17.9	29	15.9	10.8	PRESENT	PRESENT			PRESENT		0.8	81.6	5.7			BETA THALASSEMIA TRAIT
348	910999	288/2020	22	M	LUDHIANA	JAINSM	NO	NO	NO			ABSENT	6.47	12	63	18.5	29.5	16.8	9.73	PRESENT	PRESENT			PRESENT		0.8	82	5.5			BETA THALASSEMIA TRAIT
349	919320	292/2020	26	F	SANGRUR	SIKH	YES	NO	NO				3.97	10.6	86.2	26.6	30.9	13.5	21.71		PRESENT				PRESENT	0.8	53.1	2.2	35.6		HbD PUNJAB HETEROZYGOUS
350	451236	299/2020	2	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.4	10.4	63	18.8	29.9	16.8	11.66	PRESENT	PRESENT			PRESENT		2.5	80.9	4.4			BETA THALASSEMIA TRAIT
351	777161	301/2020	24	F	SANGRUR	SIKH	YES	NO	YES			ABSENT	6.2	11.6	61	18.7	30.5	16.5	9.83	PRESENT	PRESENT			PRESENT		0.9	80.7	5.8			BETA THALASSEMIA TRAIT
352	920816	308/2020	29	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.23	8.9	54.7	16.9	31	23.2	10.45	PRESENT	PRESENT			PRESENT		0.8	82.5	5.5			BETA THALASSEMIA TRAIT
353	921349	320/2020	27	F	LUDHIANA	SIKH	NO	NO	NO			PRESENT	4.54	9.1	66	20	30.3	16.7	14.53	PRESENT	PRESENT			PRESENT		1.4	82.2	5			BETA THALASSEMIA TRAIT
354	921811	332/2020	26	F	LUDHIANA	SIKH	NO	NO	NO			ABSENT	5.62	12.3	70	21.9	31.2	15.1	12.45	PRESENT	PRESENT			PRESENT		1.4	79.9	6.8			BETA THALASSEMIA TRAIT
355	921825	334/2020	26	M	MOGA	SIKH	YES	NO	NO			ABSENT	6.82	12.5	62	18.3	29.7	15.3	9.09	PRESENT	PRESENT			PRESENT		0.8	82.8	5.3			BETA THALASSEMIA TRAIT
356	922047	335/2020	31	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.09	10	63.5	19.6	31	14.9	12.47	PRESENT	PRESENT			PRESENT		0.8	82.6	5.4			BETA THALASSEMIA TRAIT
357	923653	358/2020	26	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	3.92	10.8	84	27.5	32.8	14.4	21.42	PRESENT	PRESENT			PRESENT		0.8	81.2	4.1			BETA THALASSEMIA TRAIT
358	923652	359/2020	26	M	LUDHIANA	SIKH	NO	NO	NO			ABSENT	6.96	13.4	64.1	19.3	30.1	16.1	9.2	PRESENT	PRESENT			PRESENT		0.8	79.5	6.5			BETA THALASSEMIA TRAIT
359	924840	364/2020	30	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	5.89	10.9	60.7	18.4	30.4	16.5	10.3	PRESENT	PRESENT			PRESENT		0.8	80.7	6.6			BETA THALASSEMIA TRAIT
360	924839	366/2020	32	M	LUDHIANA	HINDU	YES	NO	NO			PRESENT	5.93	11.5	63.3	19.5	30.8	15.5	10.67	PRESENT	PRESENT			PRESENT		0.8	81	6.4			BETA THALASSEMIA TRAIT
361	8264	367/2020	35	F	LUDHIANA	HINDU	NO	NO	NO			PRESENT	4.55	12	82	26.3	32	15.5	18.02		PRESENT				PRESENT	0	82.9	4.8			BETA THALASSEMIA TRAIT
362	924794	370/2020	1	M	NAWANSHAHAR	HINDU	YES	YES	NO	INCREASED	INCREASED	PRESENT	1.32	2.9	71.5	21.9	30.6	34.4	54.16	PRESENT	PRESENT					57.4	32.5	2.8			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
363	925345	375/2020	28	F	LUDHIANA	HINDU	NO	NO	NO				4.05	12.7	96.1	31.3	32.6	15.2	23.72		PRESENT				PRESENT		84.7	3.9			BORDERLINE HbA2 LEVELS SEEN
364	698207	376/2020	38	M	LUDHIANA	HINDU	NO	NO	NO				5.49	15.9	86.7	28.9	33.3	16.2	15.79		PRESENT				PRESENT		84.5	3.7			BORDERLINE HbA2 LEVELS SEEN
365	731683	379/2020	24	F	LUDHIANA	HINDU	NO	NO	NO				4.52	12.4	85	27.3	32.3	15.2	18.8		PRESENT				PRESENT	0.8	84.9	3.9			MILDLY RAISED HbA2 LEVELS NOTED
366	551197	380/2020	37	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	2.81	9	100	32.1	32.2	17.1	35.59	PRESENT	PRESENT			PRESENT		0.8	86.6	4.1			SUGGESTIVE OF BETA THALASSEMIA TRAIT
367	926649	388/2020	38	F	LUDHIANA	HINDU	YES	NO	NO				4.69	8.4	61	18	29.5	19.5	13	PRESENT	PRESENT			PRESENT		0	55.9	3.2	31.5		HbD PUNJAB HETEROZYGOUS
368	7856	393/2020	31	F	LUDHIANA	HINDU	NO	NO	NO			ABSENT	5.63	11	64.8	19.5	30.2	15.9	11.5	PRESENT	PRESENT			PRESENT		0.8	82.8	6.1			BETA THALASSEMIA TRAIT
369	8006	396/2020	37	F	LUDHIANA	SIKH	NO	NO	NO				4.45	12.7	87	28.6	32.7	16	19.55		PRESENT				PRESENT	0.8	86.1	3.8			MILDLY RAISED HbA2 LEVELS NOTED
370	38651	399/2020	65	M	LUDHIANA	HINDU	YES	NO	NO			ABSENT	4.75	9.3	66	19.7	30	16.8	13.89	PRESENT	PRESENT			PRESENT		1.5	80.7	7.3			BETA THALASSEMIA TRAIT
371	848304	427/2020	29	M	LUDHIANA	HINDU	NO	NO	NO				5.04	15	91	29.8	32.8	16.1	18.05		PRESENT				PRESENT	0.8	86.3	3.7			BORDERLINE HbA2 LEVELS SEEN
372	932189	430/2020	21	F	SANGRUR	MUSLIM	NO	NO	NO				4.9	12.8	82.6	26	31.5	17.5	16.85		PRESENT				PRESENT	0.8	85	3.5	36		HBD PUNJAB TRAIT
373	929235	434/2020	30	F	MOGA	SIKH	YES	NO	NO			ABSENT	5.56	10.5	62	18.8	30.6	16.1	11.15	PRESENT	PRESENT			PRESENT		1.6	81.6	7.1			BETA THALASSEMIA TRAIT
374	676570	449/2020	23	F	LUDHIANA	HINDU	YES	NO	NO			ABSENT	3.45	7.1	68	20.7	30.5	18.2	19.42	PRESENT	PRESENT			PRESENT		1.9	82.6	5.6			BETA THALASSEMIA TRAIT
375	934677	450/2020	29	F	LUDHIANA	SIKH	YES	NO	NO			ABSENT	4.16	7.9	61.6	19.1	31	20.8	14.8	PRESENT	PRESENT			PRESENT		0.8	82.8	5.2			BETA THALASSEMIA TRAIT
376	693856	460/2020	64	M	JALANDHAR	HINDU	YES	NO	NO			ABSENT	5.55	10.5	63	18.9	29.8	15.7	11.35	PRESENT	PRESENT			PRESENT		0.9	80.9	6.2			BETA THALASSEMIA TRAIT
377	773078	4257/2019	2	F	HOSHAIRPUR	HINDU	YES	NO	YES	INCREASED	INCREASED	PRESENT	2.48	6.4	67	24.1	35.8	31.7	27.01	PRESENT				PRESENT		93.3	3.8	1.9			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
378	479655	1624/2018	6	F	KAPURTHALA	SIKH	YES	YES	YES	INCREASED	INCREASED	PRESENT	3.8	5.2	69.1	21.6	31.3	31.3	23	PRESENT	PRESENT			PRESENT		87.7	8.9	3.4			SUGGESTIVE OF BETA THALASSEMIA HOMOZYGOUS
379	481695	1661/2018	30	F	LUDHIANA	SIKH	YES	NO	NO			PRESENT	4.84	10.6	70.4	22	31.2	17.6	14.54	PRESENT	PRESENT			PRESENT		0.7	92.7	6.6			BETA THALASSEMIA TRAIT
380			22	M	LUDHIANA	HINDU	NO	NO	NO			PRESENT	5.88	11.5	71.8	19.7	30	18.9	12.21	PRESENT				PRESENT		0.8	85.6	4.6			BETA THALASSEMIA TRAIT
381			19	F		HINDU	YES	NO	NO			PRESENT	5.5	10.5	70.9	19.1	30.6	20	12.89	PRESENT	PRESENT					0.8	84.9	5.2			BETA THALASSEMIA TRAIT
382			0.666666667	F	LUDHIANA	SIKH	YES	YES	YES	INCREASED	INCREASED	PRESENT	1.32	9	67.5	21.9	30.6	20.4	51.13	PRESENT				PRESENT		94.3	2.7	3.1			BETA THALASSEMIA HOMOZYGOUS
383			27	M		SIKH	YES	NO	NO			PRESENT	5.58	10	64.6	31.2	30.8	15.8	11.57					PRESENT		0.7	91.8	5.4			BETA THALASSEMIA TRAIT
384			24	F		SIKH	YES	NO	NO			PRESENT	5.11	10	64	20	30.2	15.3	12.52	PRESENT				PRESENT		0.5	92.4	6.3			BETA THALASSEMIA TRAIT
385			3	M		HINDU	YES	YES	NO	INCREASED	INCREASED	PRESENT	2.28	7.9	63	24	26	26	29.31	PRESENT				PRESENT		47.7	53.7	2.6			BETA THALASSEMIA HOMOZYGOUS

S.NO	MRD NO.	IMMUNOPATH NO.	AGE	SEX	PLACE OF BIRTH	CASTE/RELIGION	PALLOR	BLOOD TRANSFUSION RECEIVED	JAUNDICE	LIVER SPAN	SPLEEN SPAN	FAMILY HISTORY	RBC	Hb	MCV	MCH	MCHC	RDW	MENTZER INDEX	MICROCYTES	NORMOCYTES	PENCIL CELLS	TARGET CELLS	HYPOCHROMIA	NORMOCHROMIA	HbF	HbA	HbA2	HbD	Hb S	IMPRESSION
386			34	M			YES	YES	NO			PRESENT	5.52	11	66	30.2	30.6	15	12.7	PRESENT				PRESENT		2	92.2	5.8			BETA THALASSEMIA TRAIT
387			32	F			YES	NO	NO			PRESENT	5	7.1	66	22.7	30.5	16	13.2	PRESENT				PRESENT		1.8	91.9	5.6			BETA THALASSEMIA TRAIT
388			35	M	LUDHIANA	HINDU	YES	NO	NO			PRESENT	4.9	9.9	67	30.6	30.2	16	13.67	PRESENT				PRESENT		0.8	93.2	6			BETA THALASSEMIA TRAIT
389			32	F	LUDHIANA	HINDU	YES	NO	NO			PRESENT	4.9	10.4	66	25	30.8	15	13.46	PRESENT				PRESENT		1.1	94.4	4.5			BETA THALASSEMIA TRAIT
390			14	F	LUDHIANA	HINDU	YES	NO	NO			PRESENT	4.7	10	64	26	30.8	16	13.61	PRESENT				PRESENT		0.1	94.9	5			BETA THALASSEMIA TRAIT
391			22	F	MOGA	SIKH	YES	NO	NO			PRESENT	5.2	9	67.5	30.6	30.8	16	12.98	PRESENT				PRESENT		0.5	93	6.5			BETA THALASSEMIA TRAIT
392			34	M	FAZILKA	SIKH	NO	NO	NO			PRESENT	6.69	12.5	68.9	18.7	30.6	18.6	10.29	PRESENT				PRESENT		0.8	83.7	6.3			BETA THALASSEMIA TRAIT
393			34	F			YES	NO	NO			PRESENT	5.37	10.8	71.7	20.1	29.4	18.3	13.35	PRESENT				PRESENT		2.4	82.7	5.1			BETA THALASSEMIA TRAIT
394			0.666666667	M	LUDHIANA	HINDU	YES	YES	NO	INCREASED	INCREASED	PRESENT	3.3	6.2	79.2	25	29.2	19	24	PRESENT				PRESENT		28.2	70.4	2.6			BETA THALASSEMIA HOMOZYGOUS
395			32	M	LUDHIANA	HINDU	YES	NO	NO			PRESENT	3.23	10	66	30.6	31.2	15.3	12.61	PRESENT				PRESENT		1	93.7	4.8			BETA THALASSEMIA TRAIT
396			28	F	LUDHIANA	HINDU	YES	NO	NO			PRESENT	5.2	10	66	29.2	31.2	18	12.7	PRESENT				PRESENT		1.3	93.1	5.1			BETA THALASSEMIA TRAIT
397			1	M		HINDU	YES	NO	NO	INCREASED	INCREASED	PRESENT	3.21	9	74	30.6	29.6	22	14.2	PRESENT				PRESENT		32.2	67.1	2.5			BETA THALASSEMIA HOMOZYGOUS
398			25	M		HINDU	YES	NO	NO			PRESENT	5.2	10	66	29.2	30.6	16	12.69	PRESENT				PRESENT		1.9	92.4	3.8			BETA THALASSEMIA TRAIT
399			25	F		HINDU	YES	YES	NO			PRESENT	4.7	10	66	29.2	29.2	18	14.04	PRESENT				PRESENT		0.9	94.1	5.1			BETA THALASSEMIA TRAIT
400			1	M		HINDU	YES	YES	NO	INCREASED	INCREASED	PRESENT	4.6	9	74	29.2	30.2	28	16.08	PRESENT				PRESENT		96.4	0.4	3.2			BETA THALASSEMIA HOMOZYGOUS
401			29	M		HINDU	YES	NO	NO			PRESENT	5.2	10	66	29.2	30.2	15	12.69	PRESENT				PRESENT		0.8	93.6	3.8			BETA THALASSEMIA TRAIT
402			41	F		HINDU	YES	NO	NO			PRESENT	4.9	9.5	67	24.2	30.6	16	13.67	PRESENT				PRESENT		0.9	93.6	5.4			BETA THALASSEMIA TRAIT
403			0.25	F		SIKH	YES	YES	NO	INCREASED	INCREASED	PRESENT	2.3	5.7	77	29.3	30.6	30.6	33.4	PRESENT				PRESENT		97	0.4	1.2			BETA THALASSEMIA HOMOZYGOUS
404			26	M		SIKH	NO	NO	NO			PRESENT	5.93	13	71.6	21.9	30	20.1	12.91	PRESENT				PRESENT		0.8	82.4	5.9			BETA THALASSEMIA TRAIT
405			20	F		SIKH	YES	YES	NO			PRESENT	4.6	9.6	70.2	20.9	30	17.6	15.26	PRESENT				PRESENT		1.4	83.5	5.5			BETA THALASSEMIA TRAIT

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No. BFUHS/2K19p-TH/ 12516 Date 8/9/19

The Principal,
DMC, Ludhiana.

Subject: **Approval of Plan of Thesis.**

This is to inform you that the Faculty of Medical Sciences in its meeting held on 29.05.2019 has approved the plan of thesis of **Anshul Jain**, a student of your college for **MD Pathology** along with the names of Supervisor/Co-Supervisor as under:-

Topic Plan of Thesis

Spectrum of thalassemia and hemoglobinopathies in a tertiary care hospital

Supervisor
Dr. Bhavna Malhotra
Prof.,
Dept. of Pathology,
DMCH, Ludhiana.

Co-Supervisor
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The student has to submit the four copies of thesis (along with PDF format) , to the Controller of Examinations, BFUHS, Faridkot, at least six months before the commencement of theory examination, if he/she is otherwise eligible to take the examination. He /She must directed to attach a photocopy of this letter with the forwarding letter while submitting thesis to the University along with the letter of approval of University for change of supervisor/Co-Supervisor, change of topic,(if any).

Dr. S P Singh MS(Oph)
Controller of Examinations

Note :

1. *If any discrepancy found regarding topic of thesis or Supervisor/Co-Supervisor, inform undersigned within one month after issue of this letter.*
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SPECTRUM OF THALASSEMIA AND HEMOGLOBINOPATHIES IN A TERTIARY CARE HOSPITAL

THESIS PROPOSAL

FOR APPROVAL OF SUBJECT OF THESIS TO BE SUBMITTED IN

PARTIAL FULFILMENT OF THE REQUIREMENTS

FOR THE DEGREE OF

MD

In

PATHOLOGY

OF THE

BABA FARID UNIVERSITY OF HEALTH SCIENCES

FARIDKOT

SEPTEMBER, 2018

DR. ANSHUL JAIN

DEPARTMENT OF PATHOLOGY

DAYANAND MEDICAL COLLEGE AND HOSPITAL

LUDHIANA

ABSTRACT OF PLAN OF THESIS

Title	SPECTRUM OF THALASSEMIA AND HEMOGLOBINOPATHIES IN A TERTIARY CARE HOSPITAL
For the degree of	MD (PATHOLOGY)
Name of the candidate	Dr. ANSHUL JAIN
Supervisor	Dr. Bhavna Malhotra, Professor, Department of Pathology, DMCH, Ludhiana
Co-supervisor(s)	Dr. Vikram Narang, Assistant Professor, Department of Pathology, DMCH, Ludhiana
Institution	Dayanand Medical College and Hospital, Ludhiana
University	Baba Farid University of Health Sciences, Faridkot

Introduction: This study will be conducted for 2 years and 6 months (1 year retrospective and 1 year and 6 months prospective) in the Department of Pathology, Dayanand Medical College and Hospital, Ludhiana. All blood samples received in immunopathology lab for High Performance Liquid Chromatograhy will be analysed and correlated clinically.

Aim of Study: To analyze Thalassemia and Hemoglobinopathies at our centre.

Materials & Methods: All blood samples received in Immunopathology lab for High Performance Liquid Chromatography will be analysed.

Clinical Significance: To obtain a reasonable good database for more accurate assessment and understanding of spectrum of Thalassemia and hemoglobinopathies

Keywords: HPLC, Thalassemia, Hemoglobinopathies.

Anshul Jain		Bhavna	
(Signature)	(Signature)	(Signature)	(Signature)
Candidate	Co-supervisor	Supervisor	Principal

CERTIFICATE OF FACILITIES AVAILABLE

This is to certify that the facilities for work on the subject of thesis titled **"SPECTRUM OF THALASSEMIA AND HEMOGLOBINOPATHIES IN A TERTIARY CARE HOSPITAL"** exist in the Department of Pathology, DMC&H, Ludhiana and will be provided to the candidate. We will see that the data being included in the thesis is genuine and is collected by the candidate himself under our supervision and guidance.

Supervisor

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Place : Ludhiana

Date : 11/12/18

APPROVAL PROFORMA
BY
RESEARCH & ETHICAL COMMITTEE
DAYANAND MEDICAL COLLEGE & HOSPITAL, LUDHIANA

Name of candidate	Anshul Jain
Department	Pathology
Topic of Thesis	SPECTRUM OF THALASSEMIA AND HEMOGLOBINOPATHIES IN A TERTIARY CARE HOSPITAL
Likely date of appearing in PG examination	May 2021
Date of enrollment	10th May, 2018
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1. HOD Pharmacology	1. HOD Physiology
2. HOD Surgery	2. HOD Pathology
3. HOD Pathology	3. HOD Pharmacology
4. HOD Medicine	4. HOD SPM
5. Dean Academics	5. HOD Medicine
6. Vice Principal	6. HOD Surgery
	7. HOD Obst. & Gynae
	8. Legal Expert
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	12. Principal

Approved : Yes / No

Chairperson
Research Committee

Chairperson
Research Committee

Approved : Yes / No

Chairperson
Ethics Committee

Chairperson
Ethics Committee

BABA FARID UNIVERSITY OF HEALTH SCIENCES, FARIDKOT

PROFORMA FOR REGISTRATION OF SUBJECT FOR DISSERTATION

1	NAME OF THE CANDIDATE AND ADDRESS	Dr. Anshul Jain Ward No-5, Bhawan Kumar Jain Lambi Gali, Mukerian, District-Hoshiarpur (Punjab)
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3	COURSE OF STUDY AND SUBJECT	MD PATHOLOGY
4	DATE OF ADMISSION OF COURSE	10.05.2018
5	TITLE OF THE TOPIC	SPECTRUM OF THALASSEMIA AND HEMOGLOBINOPATHIES IN A TERTIARY CARE HOSPITAL

6. BRIEF RESUME OF THE INTENDED WORK

6.1 NEED FOR THE STUDY

Thalassemia and Hemoglobinopathies are one of the most common inherited genetic abnormalities reported worldwideparticularly in a broad belt from Mediterranean basin through the Middle East, Indian subcontinent, Burma, Southeast Asia, Melanesia and the islands of the pacific. According to Hereditary Disease Program of the World Health Organization (WHO), the carriers of haemoglobin disorders in the world are estimated to be 269 million which constitutes 4.5% of the world population.[1]

The cumulative gene frequency of hemoglobinopathies in India is 4.2% with population of 1.25 billion The carrier rate for beta thalassemia gene ranging from 1 to 3% in Southern India and 3 to 15% in Northern India [2] which means on an average 1 in every 25 Indian is a carrier of thalassemia.In India 10,000 children every year are born suffering from thalassemia major, amounting to approximately 10% of total world numbers.[3]

According to study regarding the global health burden of hemoglobinopathies, by David Weatherall, the most important forms of beta thalassemia are beta thalassemia major and haemoglobin E (Hb E) beta thalassemia, which occurs at ahigh frequency in parts of the Indian subcontinent. Haemoglobin D-Punjab is one of the most commonly encountered abnormal haemoglobins in the worldand occurs in large number of people in North West India and has a high frequency in Punjab with incidence of 2-3%.[4]

The extremely high frequency of the haemoglobin disorders compared to other monogenic diseases reflects other different processes. First, and foremost, is natural selection in which carriers have been protected during

evolution from severe malaria. Hence they have lived longer in malarious areas and produced more children until the numbers are balanced by the loss from the community of homozygotes. A second critical factor is the high number of consanguineous marriages which tend to increase the frequency of any Mendelian recessive disorder. Additional factors that maintain the high gene frequency include founder effects and gene drift.[5]

Thalassemia major is a major health problem pose economical and psychological burden on the affected individual and his family and country as a whole and needs regular blood transfusions, widespread medical care. Hence a detailed study of ethnic and geographic origin needs to be conducted for diagnosis and screening.Steps should be taken to improve the quality of life of these patients and to prevent it by population screening, genetic counselling and prenatal diagnosis and premarital counselling as well.

Almost 10 billion rupees per year is spent for thalassemia patients in India. In India due to sociocultural practices, these disorders are restricted to particular geographic areas, tribes and religion early marriages among individuals of the same caste or in close blood relations makes it to know the spectrum of thalassemia and other hemoglobinopathies. All it takes is a simple blood test to identify silent carriers so as to avoid marriages between them.[6]

6.2 REVIEW OF LITERATURE

Hereditary disorders of haemoglobin result either from qualitative defect (structural alteration of a globin polypeptide chain) due to mutations known as hemoglobinopathieswhich are HbLepore, Hb D andHb-E or due toquantitative defect(reduced or absent synthesis of one or more of normal globin polypeptide chain) known as thalassemiaswhich are alpha and beta thalassemia.[7]

Each individual receives one or more genes from each parent for each of the four major haemoglobin chains. The synthesis of beta and delta is under the control of single genes, but the gene loci of alpha and gamma chains are duplicated[8] Depending on the chains involved they are classified as alpha, beta, delta and gamma chain variants. There are two main groups of thalassemia, one affecting the synthesis of alpha chains, and the other affecting the synthesis of beta chains called as alpha thalassemia and beta thalassemia respectively. The alpha thalassemia is primarily due to gene deletion directly causing reduced alpha globin chain synthesis. Beta thalassemia is usually caused by point mutation rather than large deletion.

These subgroups have in common an imbalanced globin synthesis, with the consequence that the globin produced in excess is responsible for ineffective erythropoiesis (intramedullary destruction of erythroid precursors) and hemolysis(peripheral destruction of red cells). The mutant gene is situated at the same chromosomal locus as(i.e.is an allele of) the normal gene controlling the normal chain. The alpha and beta chains of haemoglobin are synthesised independently under separate genetic control and in the normal state, synthesis of the two chains is balanced.[9]

It is important to appreciate that great majority of abnormal haemoglobins confer no harmful effects and the individual remains asymptomatic and unaware of the abnormality within the red cell. However the majority of abnormal haemoglobins differ from the corresponding normal haemoglobin by the substitution of a single amino acid in one of their pairs of polypeptide chains. A small number have double amino acid substitutions and other have deletion of amino acids.[8]

The inheritance of thalassemia is co-dominant, and follows classic mendelian principles. Alpha thalassemia and beta thalassemia exist in both the homozygous and heterozygous states, and the genes for thalassemia may interact with those of the haemoglobin structural variants.[8]

Beta thalassemia is clinically more significant in India and is most common in communities such as Sindhi, Parsee, Lohar, and Muslims and different ethnic groups of Punjab, Bengal, bhanushalis, kutchis, lohanas from Gujarat[9], mahars, neobuddhists, kolis, and agris from Maharashtra and gowdas and lingayats from Karnataka etc. Concurrent presence of alpha thalassemia modifies the phenotype of beta thalassemia. Alpha thalassemia is not a significant problem in India because the severe alpha deletion mutations are less common in this region and is clinically silent but is widely prevalent in tribal population.

Table 1 explaining various Thalassemia syndromes[10]

CLINICAL SYNDROMES	GENOTYPES	CLINICAL FEATURES
β thalassemias		
β thalassemia major	(β^0/β^0, β^+/β^+, β^0/β^+)	Severe; Blood transfusion dependent
β thalassemia intermedia	(β^0/β^+, β^+/β^+, β^0/β, β^+/β)	Severe but does not require regular blood transfusions
β thalassemia minor	(β0/β, β+/β)	Asymptomatic with mild or absent anemia; red cell abnormalities seen
α thalassemias		
Silent carrier	-/α α/α	Asymptomatic no red cell abnormality
α thalassemia trait	-/-α/α -/α-/α	Asymptomatic red cell abnormalities seen like β thalassemia minor
HbH disease	-/- -/α	Severe; resembles β thalassemia intermedia
Hydrops fetalis	-/- -/-	Lethal in utero without transfusions

In beta thalassemia, the inadequate production of beta chains leads to reduction in the amount of Hb-A in the red cell and a microcytic hypochromia results but the total haemoglobin is maintained in part by the production of gamma and beta chains and thus increased Hb-F or Hb-A2 is usually found. In alpha thalassemia the levels of Hb-A, Hb-F, and Hb-A2 are equally depressed since they all have alpha chains, there is usually a microcytic hypochromia. Therefore in absence of sufficient alpha chains excess beta and gamma chains aggregate to form Hb-H (beta4) or Hb-Bart's (gamma4).

The pathophysiology of the thalassemias can be traced to the deleterious effects of the excessively produced globin-chain subunits. In β-thalassemia, excess chains damage the red cell precursors and red cells, leading to profound anemia. The result is extensive expansion of erythropoietic marrow, which is ineffective in producing mature red cells but impinges on developing bones, severely affecting development, bone formation, and growth. The major cause of morbidity and mortality is the effect of iron deposition on the endocrine organs, liver, and heart, which results from increased intestinal absorption and the effects of blood transfusion.[11]

The pathophysiology of the α-thalassemias is different because the excess β chains resulting from defective alpha-chain production form β4 molecules, or hemoglobin H, which is soluble. However, hemoglobin H is unstable and precipitates in older red cells. Hence, the anemia of alpha-thalassemia is hemolytic rather than dyserythropoietic.[12]

Because the structural hemoglobin variants and the thalassemias occur at a high frequency in some populations, the two types of genetic defect can be found in the same individual and can produce important clinical

manifestations. These are known collectively as the *thalassemia syndromes*. Other hemoglobin molecules which are widely prevalent in India include Hb S, Hb E and Hb D. HbE is widely distributed in the North-Eastern states of India.[13]It arises from the substitution of lysine for glutamic acid in the 26th position of the beta chain. HbD is predominantly seen in Punjab, Uttar Pradesh, Gujarat, Jammu & Kashmir. Hb D arises from the substitution of glutamine for glutamic acid in the 121st position of beta chain Hb D in both heterozygous and homozygous form is clinically asymptomatic. Other rare types of thalassemic disorders and Hb variants sporadically reported from India include δβ thalassemia, Hereditary persistence of fetal hemoglobin, Hb Lepore, Hb Q, Hb K, Hb J, Hb M, Hb Chandigarh, Hb Koyadora etc.[14]

Several studies were carried out in India in patients diagnosed to have thalassemias.

In a one-year study, in Apollo Hospitals, Chennai, a tertiary care hospital in South India, conducted on 543 patients, the commonest disorder was β-thalassemia trait (37.9%)followed by HbE trait (23.2%), homozygous HbE (18.9%), HbS trait(5.3%), HbE β-thalassemia (4.6%), HbS β-thalassemia (2.5%), β-thalassemia major(2.3%), HbH(1.6%), homozygous HbS(1.4%), HbD trait(0.7). Of these 53.2% were males and 46.7% females. On comparing the thalassemic disorders (β-thalassemia trait, β-thalassemia major, and α-thalassemias), it was observed that the lowest average hemoglobin was seen in β-thalassemia major. The lowest average MCV was seen in β-thalassemia trait, whereas the lowest average MCH and MCHC in α-thalassemias. The average Hb F was 88% in β-thalassemia major and less than one in α-thalassemias (Hb H).

Children suffering from beta-thalassemia major require repeated blood transfusions which may be associated with dangers like iron overload and contraction of infections such as HIV, HCV, and HBsAg which ultimately curtail their life span. On the other hand, inadequate transfusions lead to severe anemia and general fatigue and debility.[15]

So thalassemias and hemoglobinopathies are significant health hazard and their prevention is paramount for the society.

6.3 AIMS AND OBJECTIVES OF THE STUDY

To Study Spectrum of Thalassemia and Hemoglobinopathies.

7. MATERIALS AND METHODS

7.1 SOURCE OF DATA

The present descriptive study will be total for the period of 2.5 years (1 year retrospective and 1.5 years prospective) which will be conducted in the Department of Pathology, Dayanand Medical College and Hospital, Ludhiana. All blood samples received in the immunopathology lab for High Performance Liquid Chromatography will be analysed.

7.2 METHOD OF COLLECTION OF DATA

Inclusion criteria

All blood samples which will be coming for HPLC.

Exclusion criteria

None

Methodology

1) Clinical history, examination and investigations of all identified cases of thalassemia and hemoglobinopathies will be done as per proforma.

2) Anticoagulated blood using 3ml EDTA vial will be collected and will be run and processed in automated hematology analyzer for complete blood count. Automated analyzers are Coulter LH 750 Analyzer, UnicelDXH 800 Coulter cellular Analysis System and Horiba ABX Micros ES 60.

The instrument works on the principle of Electrical impedance (Coulter principle). Cells are sized and counted by detecting and measuring changes in electrical resistance when a particle passes through a small aperture. Height of the pulse generated is proportional to cell volume (size) and the number of pulses indicates cell number (count). Hb, TLC, DLC, Red cell

indices, mentzer's index and platelets are analyzed using automated hematology analyzer.

3) Peripheral smear will be studied after staining with Giemsa stain or other stains whenever required.

4) High Performance Liquid Chromatography is performed with each anticoagulated blood sample which is coming as 3ml EDTA vial in Biorad variant D-10. HPLC is the method of choice for initial screening of Hb variants like Hb S, Hb D, Hb E, Hb C and for accurate quantification of Hb A Hb F and Hb A2 concentrations. Each sample taking only 6.5 minutes for analysis.

A dual-piston, low pulsation High Performance Liquid Chromatography pump and a proportionating valve deliver the buffer solution to the analytical cartridge and detector. Whole blood samples undergo an automatic two-step dilution process and are introduced into the analytical flow path. Prediluted samples are identified based upon the use of a sample vial adapter in the sample rack, and the automatic dilution step is omitted. Pre diluted samples are aspirated directly and introduced into the analytical flow path. Between sample injections, the sample probe is rinsed with Wash/Diluent Solution to minimize sample carryover.

A Programmed phosphate ion gradient generated by mixing 2 buffers of increasing ionic strength delivers the sample to the analytical cartridge, where the different hemoglobins are separated based upon their ionic interactions with the cartridge material. The separated hemoglobins then pass through the photometer flow cell where changes in absorbance are measured at 415nm. A secondary filter at 690 nm corrects the effects caused by mixing buffers of different ionic strengths.

The software performs a reduction of raw data collected from each analysis that may include use of a calibration factor. A sample report and chromatogram where the different peaks are identified in defined windows with relevant information like retention time, relative percentage and area.

Study Design

This will be a descriptive study total for the period of 2.5 years (1 year retrospective and 1.5 years prospective) which will be conducted in the Department of Pathology, Dayanand Medical College and Hospital, Ludhiana

Outcome

To evaluate the spectrum of thalassemia and hemoglobinopathies using HbA_2 etc in a tertiary care hospital.

Statistical Analysis

Data will be described in terms of range; mean ±standard deviation (± SD), median, frequencies (number of cases) and relative frequencies (percentages) as appropriate. All statistical calculations will done using SPSS (Statistical Package for the Social Science) SPSS 21 version statistical program for Microsoft Windows.

7.3 DOES THE STUDY REQUIRE ANY INVESTIGATIONS OR INTERVENTIONS TO BE CONDUCTED ON PATIENTS OR OTHER HUMANS OR ANIMALS? IF SO, PLEASE DESCRIBE BRIEFLY.

No.

7.4 HAS ETHICAL CLEARANCE BEEN OBTAINED FROM YOUR INSTITUTION IN CASE OF 7.3?

Attached herewith

LIST OF REFERENCES

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PROFORMA

MRD NO:- **WARD/OPD:-** **DATE:-**

IDENTIFICATION DATA:-

NAME:- AGE:- SEX:-

DATE OF BIRTH MOBILE NO:-

DEMOGRAPHIC DATA:-

NAME OF FATHER/HUSBAND:- AGE

NAME OF MOTHER/WIFE:- AGE

PLACE OF BIRTH:- CASTE/RELIGION:-

EATING HABITS:-

LANGUAGE:-

CLINICAL HISTORY:-

PALLOR:-YES/NO JAUNDICE:-YES/NO

GENERALIZED WEAKNESS:-YES/NO

PAIN ABDOMEN:-YES/NO HEMOLYTIC FACIES:-YES/NO

FATIGUIBILITY:-YES/NO

BLOOD TRANSFUSION RECEIVED:-YES/NO NO OF UNITS

DATE

OTHERS

PHYSICAL EXAMINATION:-

LIVER SPAN

SPLEEN SPAN

FAILURE TO THRIVE/GROWTH RETARDATION

OTHERS

FAMILY HISTORY (IFANY):-

RELEVANT LABORATORY FINDING (INVESTIGATIONS):-

Signature of Candidate **Signature of Supervisor**

HEMATOLOGICAL PARAMETERS:-

TLC	RBC	Hb
PLATELET COUNT	DLC	RED CELL INDICES
MCV	MCH	MCHC
RDW	PCV	RETICULOCYTE COUNT
MENTZER INDEX		OTHERS

PBF FINDINGS:-

NORMOCYTIC	MICROCYTIC	MACROCYTIC
HYPOCHROMIA	NORMOCHROMIA	
TARGET CELLS	ELLIPTICAL CELLS	FRAGMENTED CELLS
NUCLEATED CELL		
OTHER		

HPLC AND Hb ELECROPHORESIS:- IMMUNOPATHOLOGY No:-

HbF

HbA

HbA2

HbD

HbC

HbE

IMPRESSION

REPORTS OF ANY MOLECULAR INVESTIGATION DONE:-

OTHER INVESTIGATIONS:-

SERUM IRON PROFILE

TIBC	UIBC	Transferrin saturation

SERUM FERRITIN

SERUM FOLATE

OTHER

RADIOLOGICAL PROFILE (SKULL X-RAY):-

Signature of Candidate **Signature of Supervisor**